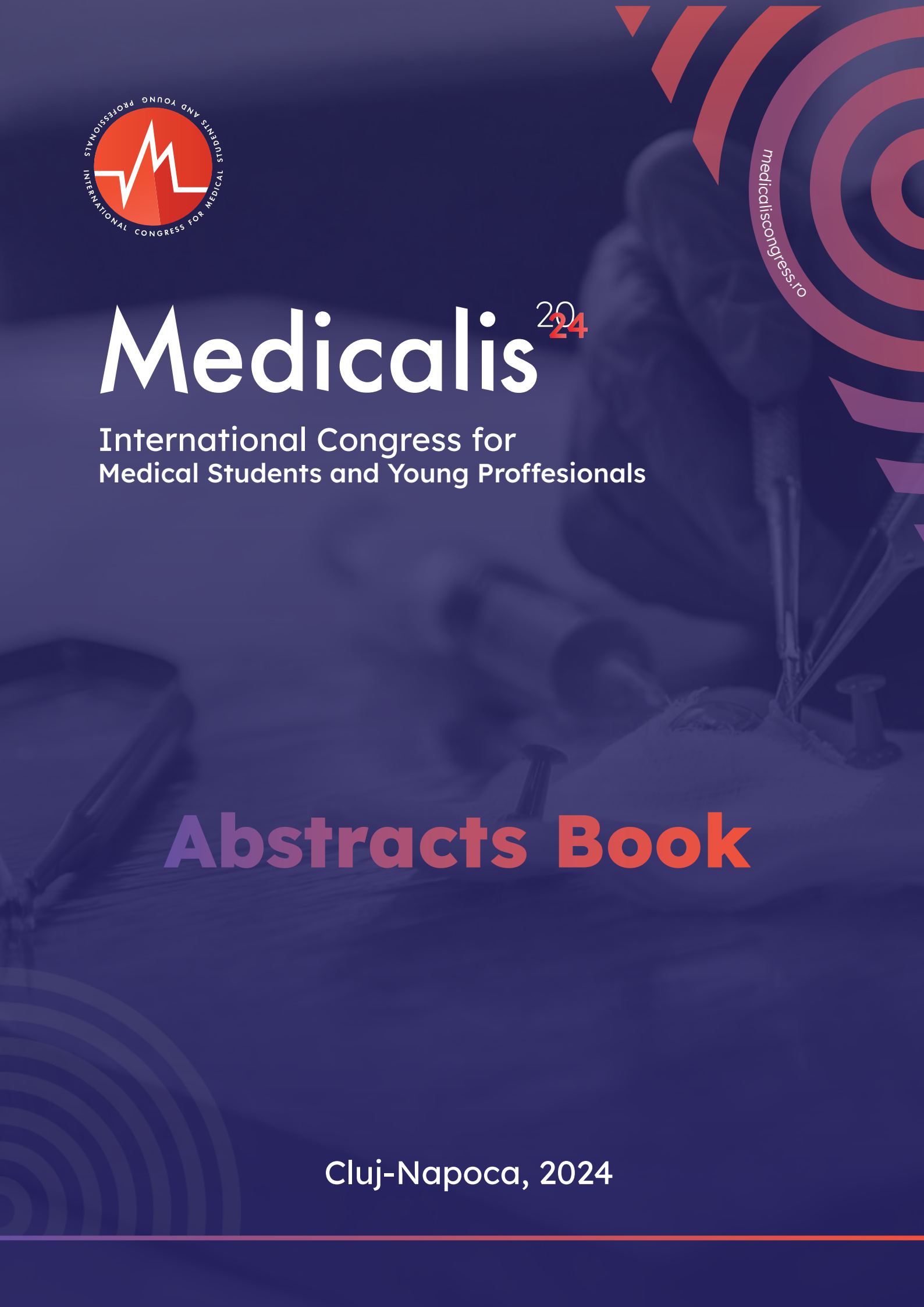
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Section 1

Oral Presentations

1.1 Winners of the competition

First Place

The Power of Nutrition - Conquering the Female Athlete Triad

Author: Daria Păpară[1]
Coordinator: Otilia-Andreea Tarcău MD[1]

Affiliations:
1-"Grigore T. Popa" University of Medicine and Pharmacy

Background
The Female Athlete Triad (FAT) combines menstrual dysfunctions (MD), poor bone health, and low energy availability (EA) which can be present with varying severity in female athletes of all ages who engage in regular, moderately intense physical exercise. Due to nutrient restriction and/or excessive energy expenditure, neuroendocrine adaptations occur to support the vital systems, interrupting ovulation and bone turnover and leading to amenorrhea, osteopenia, and increased risk of fractures.

Objectives
Transdermal hormonal therapy, combined oral contraceptives, or bisphosphonates, frequently utilized in the management of the triad's symptoms, are associated with various adverse effects and may not always address the underlying cause effectively. This review emphasizes the need for a higher EA as higher caloric intake to protect overall physiological health and prevent potentially serious consequences in female athletes, with the possibility to effectively restore fertility and enhance bone health in the majority of cases, when implemented correctly.

Material and method
For the present review, 15 relevant studies from PubMed (2018-2024) were included, highlighting low EA with/without eating disorders (ED) as the main factor underpinning hormonal disturbances that affect bone metabolism and fertility. The presence of at least one component of FAT in regularly physically active females was the inclusion criterion, ranging from subclinical menstrual disorders to functional hypothalamic amenorrhea, from low bone mass density (BMD) to osteoporosis. Patients with chronic/congenital pathologies causing amenorrhea or impairments in BMD and Z-Score were excluded.

Results
EA is crucial for a healthy state and optimal performance, typically exceeding 45 kcal/kg of fat-free mass (FFM)/day. Low EA (<30 kcal/kg FFM/day, resting metabolic rate) prioritizes survival-related processes over non-essential ones like reproduction and growth, resulting in severe chronic issues - amenorrhea and osteoporosis. Increasing body weight by 5-10% or by 1-4 kg with a nutritional surplus of 300-600 kcal/day (for at least 6 months) and reducing resistance training restores the physiological hormonal balance, promoting positive changes in menstrual regularity and BMD.

Conclusion
Nutritional therapy, the primary treatment for recovering BMD and resuming menses in energy-deficient female athletes, reestablishes the hypothalamic-pituitary-ovary axis. Given that up to 87% of female athletes experience a FAT-related disorder, dietitians should provide performance nutrition education to promote and enhance optimal EA.

Keywords: Female Athlete Triad, Energy Availability, Amenorrhea

Second Place

Who's treating me? – a multispecialty approach to Irritable Bowel Syndrome

Author: Mara-Lorena Imbrea[1]
Coordinator: Simona Ștefania Juncu MD[1, 2]

Affiliations:
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2- Institute of Gastroenterology and Hepatology, "Sf. Spiridon" University Emergency County Hospital, Iasi

Background
Irritable Bowel Syndrome (IBS) is a chronic disorder of the gut-brain interaction that affects the quality of life. With a prevalence of 1 in 10 people globally and present therapies being only modestly effective, IBS is surely becoming a socio-economic concern world-wide. This review aims to draw attention to the lack of collaboration between health professions when it comes to different management therapies of IBS.

Material and method
This review brings together the results of 11 articles between 2018 and 2023 as to signal the need of combined and personalized pharmacological and psychologic treatment to relieve symptoms. The literature search for reviews summed up to the total of 4233 patients and 118 studies. The inclusion criteria were 1) IBS patients that claim symptoms of abdominal pain at least 1 day/week in the last 3 months, change of frequency of stool (constipation, diarrhoea, or both) or change in form of stool (Rome V criteria) 2) IBS patients that have presented anxiety or depressive symptoms either before or after being diagnosed. The exclusion criteria reffered to patients diagnosed with celiac disease, Crohn's disease or ulcerohemorrhagic rectocolitis.

Results
The notable findings were: 1) the prevalence of anxiety and depressive symptoms in IBS patients being 39% and 29% respectively; 2) a dysbiosis frequency of 73% observed in IBS patients compared to a 16% rate in healthy individuals; 3) higher IBS symptom improvement in subjects receiving antidepressants (tricyclic antridepressants) compared to placebo groups (63.8%, 224/351 vs. 42.7%, 186/436 respectively); 4) brain imaging uncovering disturbed intrinsic brain activity in brain regions of IBS patients pertaining to emotional arousal; 5) a number of 83 articles illustrating the benefit of using cognitive behavioral therapy, acupuncture and diet as IBS therapy.

Conclusions
The usual clinical approach of IBS patients only includes the pharmacological alleviation of symptoms, whereas the demonstrated psychological onset calls for a non-drug approach to their symptom management. That being said, it is for the long-term benefit of the patient that the treatment includes some of the newest evidence-based therapies such as: cognitive behavioral therapy, acupuncture and mindfulness based stress reduction.

Keywords: IBS, anxiety, gastrointestinal symptom management.

Third place

Immediate breast reconstruction (IBR) versus delayed breast reconstruction (DBR) with free DIEP flap – What if we don't wait?

Author: Maria-Cristina Simian[1]
Co-author: Tabita Alexa[1]
Coordinator: Maximilian Muntean MD[1, 2]

Affiliations:
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2- Institute of Oncology "Prof dr. Ion Chiricuţă", Cluj-Napoca

Background
The gold standard for autologous flap in breast reconstruction is the Deep Inferior Epigastric artery Perforator flap (DIEP). The DIEP flap technique involves breast reconstruction using abdominal tissue transfer to the mastectomy site as a free microsurgical transplant. While older techniques involve harvesting a small amount of rectus abdominis muscle along with skin and subcutaneous tissue, the DIEP flap technique provides complete muscle preservation. Depending on the timing of the intervention, breast reconstruction can be immediate (IBR), performed in the same operation as the mastectomy, or delayed (DBR), performed months to years after the mastectomy.

Objectives
The objective of this paper is to establish whether IBR or DBR with an autologous free DIEP flap should be the preferred approach in most cases by comparing them in terms of aesthetic outcome, psychological distress, treatment costs, and associated risks.

Material and Method
We conducted research on PubMed as the preferred database. The keywords used are "Immediate DIEP flap breast reconstruction" and "Delayed DIEP flap breast reconstruction", connected by the Boolean operator "OR". The papers we took into consideration are those that were published in the last ten years (2014-2024). This search algorithm yielded 264 results.

Results
IBR implies a decreased risk of emotional distress, usually with better aesthetic results, and possibly a reduced number of surgeries with subsequent lower costs in the long term. However, IBR comes with possible skin and nipple perfusion problems and a higher occurrence of hematoma and seroma. Nevertheless, the DBR procedure more often showed wound problems. One impediment in choosing IBR might be the possible future need for radiotherapy, as it could compromise the reconstructed breast, but more research needs to be done as the indications are still unclear in this case. Furthermore, several papers claim that there is no difference in the rate of development of local cancer recurrence between the two techniques. Additionally, no significant differences were found regarding fat necrosis, partial flap loss, and total flap loss rate. It is fundamental to acknowledge that the presented complications are higher in patients who present several comorbidities, like smoking and a

higher BMI.

Conclusions
To conclude, we can state that IBR with free DIEP flap appears to be the superior intervention option after a mastectomy, but the clinician needs to keep in mind all the risk factors that might be present when deciding. Thus, the solution should be an individualized treatment plan that provides the best outcome for the patient.

Keywords: DIEP flap breast reconstruction, Immediate breast reconstruction (IBR), Delayed breast reconstruction (DBR), breast cancer, mastectomy

First Honorable Mention

Assessing Prognostic Factors for Clinical Outcomes in Malignant Melanoma

Author: Andrada Adelaida Pătrășcanu[1]
Co-authors: Sebastian-Mihai Belbe[1], Ioana-Bianca Moldovan[1], Alexandra-Iulia Păucean[1], Teodora-Maria Oros[1]
Coordinator: Assistant professor Andrada-Larisa Deac[1]

Affiliations:
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Background
Melanoma is a neoplasm developed from the pigment-producing cells, the melanocytes, being acknowledged as the fifth most common cancer worldwide due to its rapidly increasing incidence. Unlike other solid tumours, it is frequently diagnosed in people younger than 55 years old, which usually present risk factors like UV radiation exposure, family history, atypical mole syndrome, and white race. Several mutations such as BRAF, affect the MAPK pathway which is known to be involved in the development of malignant melanoma. Notwithstanding, the prognostic role of BRAF mutation is contentious, its presence seemingly linked with a worse survival outcome, but at the same time, it is considered to be one of the most immunogenic malignancies, as melanoma and its tumoral microenvironment represent an incredible target for immunotherapy and a valuable way to predict the evolution of the patients.

Objectives
To determine the influence of histopathologic features and biologic factors on immunotherapy response rate and disease prognosis.

Material and method
This assessment involved 4 patients diagnosed with stage III melanoma, two of which were BRAF positive (females) and two BRAF negative (males). The following variables were analyzed: age, gender, tumour site, neutrophil-to-lymphocyte ratio (NLR), platelet-to-lymphocyte ratio (PLR), lymphocyte-to-monocyte ratio (LMR), tumour-infiltrating lymphocyte (TIL), growth pattern, Breslow thickness, and ulceration.

Results
Our analysis identified that 75% of our patients had high Breslow index(4-22), ulcerative lesions, and non-brisk TILs. 50% presented vertical growth pattern, sole localization, low CD8 (20-40%), low FOXP3 (30-60%), and subunitary CD8/ FOXP3 ratio. NLR ranged between 2-4, PLR between 101-244, and LMR between 1,54-3,28, but they possess low specificity because of the cut-off variations. Every patient enrolled in our study has clinical prognostic factors associated with a poor prognosis, particularly the BRAF-negative patients that showed progressive disease after 3 cycles of immune checkpoint inhibitors (ICI), whereas one of the BRAF-positive patients treated with Dabrafenib and Trimetinib (D+T) had one-year progression-free survival and the other presented progressive disease after 6 cycles of ICI + ongoing D+T.

Conclusions
The improved survival of BRAF-positive patients is related to their higher CD8 and FOXP3 percentages, which make this type of melanoma a highly immunogenic tumour model. However, data are controversial and require future confirmations because of the small number of patients. In addition, we need reliable markers so as to be able to identify which patients will respond to immunotherapy.

Keywords: malignant melanoma, BRAF mutation, immune checkpoint inhibitors, B-raf inhibitors, melanoma prognostic factors

Second Honorable Mention

Human Amniotic Membrane Biological Dressings in Burn Therapy - An Accelerating Adjuvant for Standard Skin Grafts?

Author: Alexia-Gabriela Stratan [1, 2]
Co-authors: Catinca Dumitrescu[1], Mara-Georgiana Uliliuc[1], Cosmin-Ștefan Velnic[1]
Coordinator: Lecturer Camelia Tamaș MD, PhD [1, 3]

Affiliations:
1-"Grigore T. Popa" University of Medicine and Pharmacy Iași
2-Romanian Student Society of Surgery
3-"Sf. Spiridon" Emergency Clinical County Hospital Iași

Background
Burns are amongst the most traumatic skin injuries, leaving large defects which render the host susceptible to bacterial infections. Complications lead to systemic sepsis and severe septicemia (75% of mortality in burned patients), obliging for appropriate wound care management. Human amniotic membrane (HAM) is a multilayer membrane separated from the chorion and placenta. Showing low antigenicity and promising support of cell proliferation, HAM makes for an auspicious alternative to traditional burn dressing, not only regarding healing time, but also proving to decrease the tissue bacterial bioburden.

Objectives
The aim of this review is to assess the functional and clinical efficacy of sterilized HAM as an alternative dressing in burn wound care.

Material and Method
This review is based on 11 PubMed articles (10 Original Studies, 1 Meta-Analysis) published between 2011-2020, totalling 632 patients. Inclusion criteria comprised patients with second or third-degree burns (4-15% of total body surface area) requiring split-thickness skin grafts, while those presenting less than second-degree burns or history of cardiac disease, renal failure and underlying metabolic disorders were excluded. HAMs were isolated from the placentae of screened donors based on medical history, followed by succesive washing, air-drying and gamma-ray sterilization. The layer was then applied on the wound bed after split-skin were harvested and grafted, accompanied by Vaseline and dry gauze dressing.

Results
Results were proven satisfactory regarding promotion of wound healing, with a complete graft take rate of 96.76% and a mean duration of 6.98 ± 1.35 days, significantly overcoming the traditional 13.99 ± 1.6 days. HAM is accompanied by rapid re-epithelialisation and granulation-tissue development by inhibition of leucocyte protease activity and angiogenesis stimulation. Moreover, laboratory investigations revealed that the basement membranes of amnios share major components with human skin, thus resembling it both morphologically

and ultrastructurally. Procurement and processing are facile, while sterilization and preservation do not require consequential costs and show no obvious architectural changes over time. However, the main concern focuses on whether the antibacterial activity against burn-isolated resistant bacteria is potent, although it is safe to state that HAM dressings were found impermeable to various bacili and cocci strains.

Conclusions
HAM biological dressings are a safe and feasible alternative to routine treatment course in burn therapy with providential prospects for wound protection, microbial control and maturation hastening, showing strong potential for a better over-all management of these traumatic events.

Keywords: human amniotic membrane; biological dressings; burns; skin substitute

1.2. Basic Medical Science

Targeted therapies in polycystic ovary syndrome

Author: Maria Vancea[1]
Coordinator: Svetlana Capcelea[1]

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Background
Polycystic ovary syndrome (PCOS) is a complex endocrine pathology manifested by biochemical and clinical hyperandrogenia, is associated with ovulatory dysfunction and the appearance of multiple cystic formations at the ovary level. Elucidation of pathogenesis, diagnosis and treatment of PCOS are permanent concerns.

Objectives
Study of ethio-pathogenetic factors and molecular targets in the treatment of PCOS.

Materials and methods
The research is based on bibliographic analysis of the sources identified in PubMed, NCBI, Research Gate and Medline, published in 2014-2024.

Results
PCOS is a complex and highly heterogeneous etiological pathology with the combination of various genetic, epigenetic and environmental factors. In the pathogenesis of PCOS, severe biochemical pathways have been studied in which products from various genes encoding factors involved in the biosynthesis and metabolism of steroid hormones (CYP17, CYP11, CYP19, CYP19, CYP19, HSD3B1-2, HSD17B1-3, StAR), insulin secretion and action (IGF1, IGF1R, IGFBPI1-3, INS VNTR, IR, INSL, IRS1-2, PPARG), obesity and energy regulation (MC4R, OB, OBR, POMC, UCP2-3), gonadotropin and gonadal hormones action (ACTR1, ACTR2A-B, FS, INHA, INHBA-B, INHC, SHBG, LHCGR, FSHR, MADH4, AR). Different variations in these genes could lead to predisposition to anovulatory PCOS and the concomitant risk of developing type 2 diabetes. Mutations in severe aromatase-coding genes, including CYP11A1, CYP11B2, CYP17A1, CYP19A1, CYP1A1, CYP21A2, CYP3A7, have been reported to be involved in PCOS pathogenesis. These genes and/or products of their expression become biomarkers in the diagnosis of PCOS and potential therapeutic targets. Potential therapies in PCOS include exosome therapy, gene therapy, and drugs based on targeting aberrant pathways by alleviating inflammation, insulin resistance, excess androgen, and ovarian fibrosis, what are typical pathological elements of PCOS. Researchers are concerned about understanding gene-gene, gene-environment relationships when administering different biomolecules with affinity to signaling pathways responsible for the pathogenesis of PCOS: PI3K/Akt, TLR4/NF-κB, Nrf2/HO-1, AMPK, MAPK, JAK/STAT, Wnt/β-catenina, Notch, Hipo/YAP, TGF-β/Smad and hedgehog paths. In women with PCOS, an excess of LH and an inadequate FSH/LH ratio are observed, and the new therapeutic substances could base their action on GABA inhibition and stimulation of the kisspeptin of GnRH neurons. Proteins from the GLUT type 4 family are targets for new agents that would combat insulin resistance in patients with PCOS.

Conclusion
PCOS is a multifactorial pathology, determined by the varied interaction of predisposing genetic changes and environmental factors. The personalized approach of patients with PCOS involves the correlation of metabolic disorders including ovulatory dysfunction, insulin resistance, obesity with molecular-genetic tests.

Keywords: Hyperandrogenism, insulin resistance, polycystic ovary syndrome, signaling pathways, targeted therapies.

Harmonizing Recovery: Music Therapy in Traumatic Brain Injury resulting from Motorcycle Accdients with Helmet Breakage

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Co-author: Daria-Ioana Vreme[1]
Coordinator: Emilian-Bogdan Ignat Teaching Assistant MD[1]

Affiliations:
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Background
Traumatic Brain Injury (TBI) encompasses a spectrum of neurologic dysfunction resulting from external force to the head and possesses multifaceted challenges, necessitating innovative approaches beyond conventional treatments. Music therapy has emerged as a potential ally in the rehabilitation journey of TBI patients, offering holistic benefits across cognitive, emotional, and physical domains.

Objectives
This study seeks to explore the effectiveness of music therapy as a supplementary treatment approach in individuals suffering from Traumatic Brain Injury (TBI) following motorcycle accidents wherein helmet breakage has occurred.

Material and Method
Our study employed a systematic review methodology to investigate music therapy`s efficacy in traumatic brain injury (TBI) in patients from motorcycle accidents with helmet breakage. In conducting analysis, we searched across esteemed databases, including Web of Science (Core Collection), PsycINFO and Google Scholar as well as accessing articles published between 2005 - 2020 via PubMed. Initially, 5,472 studies were screened for eligibility, 228 being found suitable for encompassment in our analysis. Our inclusion criteria targeted studies investigating music`s impact on cognitive, emotional, and psychosocial aspects of TBI rehabilitation on individuals aged 18 and above, who suffered a motorcycle accident with helmet breakage. We focused on studies examining music therapy`s impact on cognitive, emotional, and psychosocial aspects of TBI recovery, considering diverse methodologies such as experimental research and clinical trials.

Results
The trial has shown advancements among patients in both physical and cognitive domains, as well as in social engagement through activities such as reading, games, music, artwork, and internet access, complemented by standard inpatient rehabilitation services. Among the 228 patients evaluated, cognitive function displayed recovery rates ranging from 25% to 75%, while emotional well-being demonstrated developmental progress in 30% to 60% of cases. Motor skill recovery was observed at rates between 40% and 70%, with variations in quality of life noted within the range of 30% to 70%.

Conclusion

Music therapy shows promise as an effective intervention for addressing the complex needs of patients with TBI resulting from motorcycle accidents with helmet breakage. By integrating music-based interventions into comprehensive rehabilitation programs, healthcare providers can enhance cognitive function, promote emotional well-being, and facilitate the overall recovery process in this vulnerable population.

Keywords: Traumatic Brain Injury, Music Therapy, Motorcycle Accidents, Helmet Breakage

Could your occupation cause skin cancer? Deciphering the connection between one face and two sides of a story

Author: Daria-Ioana Vreme[1]
Co-author: Daria-Alexia Grigoraș[1], Daria-Elena Druhuș[1], Ioana Adumitresei[1]
Coordinator: Teaching Assistant MD, PhD Irina-Mihaela Jemnoschi-Hreniuc[1]

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Background
Occupational factors play a significant role in the risk of developing skin cancer, alongside environmental exposures such as ultraviolet radiation (UVR), personal habits, and genetic predisposition. Over the last 50 years, the global incidence of skin cancer has surged dramatically, with basal cell carcinoma (BCC) emerging as the predominant form, followed by squamous cell carcinoma (SCC) and melanoma which, despite its lower prevalence, presents as the most aggressive variant, with heightened metastatic potential.

Objectives
This review aims to magnify the correlation between sun exposure on distinct facial regions and the primary types of skin cancer incited by this factor.

Material and Method
In the process of conducting this systematic review, extensive searches were conducted across multiple databases including Embase, Medline (Ovid), and Web of Science (Core Collection), alongside accessing articles on PubMed published between 2007-2023. Our inclusion criteria focused on primary research studies investigating the impact of sun exposure on individuals aged 25 and above, specifically examining the risk of the three predominant types of skin cancer. Our analysis encompassed over 1500 cases, with approximately 1300 BCC, 100 SCC and the remaining cases pertaining to melanomas. Additionally, our research indicates that certain occupations are associated with an elevated risk of skin cancer in specific facial regions. Hence, our study investigated individuals employed as taxi drivers, truck drivers, farmers, railway engine drivers and workers, firemen, miners, and quarrymen. Odds ratios (OR) and 95% confidence intervals (CI) were estimated using logistic regression mixed models.

Results
For skin cancer as a whole, miners and quarrymen registered excess risk, regardless of exposure to solar radiaton and skin type. Frequency of BCC proved higher among railway engine drivers and firemen (OR 4.55; 95% CI 0.96–21.57), and farmers (OR 1.65; 95% CI 1.05–2.59). Furthermore, research has demonstrated that railway workers exhibit a heightened susceptibility to BCC on the forehead, while taxi and truck drivers have been found to develop BCC on the left auricular helix, with SCC commonly observed on the left

arm. The occupations that registered a higher risk of SCC (though not of BCC) in those involving direct contact with livestock and construction workers (OR 2.95, 95% CI 1.12–7.74).

Conclusions

The association between occupation and skin cancer incidence, as demonstrated by distinct patterns across various professions, underscores the importance of recognizing and addressing these occupational risks. Understanding these occupational risks is crucial for implementing targeted preventive strategies to safeguard the health of workers worldwide.

Keywords: occupation, squamous cell carcinoma, basal cell carcinoma, melanoma;

1.3. Clinical Medical

The importance of spirometry in bronchial asthma management and the predictive value of MMEF and FEV1 parameters for future exacerbations

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Background
Spirometry is currently the most used and most reliable method to diagnose bronchial asthma and to follow its evolution (control assessment of symptoms and risk for exacerbations). A recent study has shown a suboptimal control of bronchial asthma and other studies showed that the prevalence of asthma is increasing. That is why we considered necessary a study that shows the correlation between spirometry parameters and symptomatology, along with the risk for future exacerbations of asthmatic patients.

Objectives
The importance of performing spirometry in bronchial asthma monitoring, correlating the values of the functional parameters with the number of exacerbations and quantifying this correlation highlighting the predictive value of FEV1 and MMEF for the risk of future exacerbations.

Material and Method
It is a cohort, observational, retrospective study. It was carried out in the Emergency Clinical Hospital for Children in Cluj-Napoca, Pediatric Clinic III. For this study, the closed circuit method of spirometry was used. The size of the entire sample was 480 patients who performed spirometry, of which 184 were diagnosed with bronchial asthma. 28 patients with asthma were randomly selected and followed up for 2 years (January 2022- December 2023)

Results
Values of the spirometric parameters FEV1 and MMEF were collected from all patients with asthma (n=184). The comparison between the values of FEV1 for patients with controlled (C) and uncontrolled (UC) symptomatology, showed lower values of FEV1 in the UC group (p=0,015). A 10% higher frequency of patients with FEV1 values <80% was observed in the NC group, compared to the C group. The number of exacerbations during two years was tracked in correlation with the values of spirometric parameters at the beginning and then at the end of the follow-up period. The regression model was statistically significant (p=0.006) for MMEF values (Spearman Multiple correlation coefficient R=0.26).

Conclusions

Patients with uncontrolled bronchial asthma presented significantly lower values of FEV1 compared to the group of patients with controlled bronchial asthma, thus reflecting the impairment of lung function at the time of spirometry, but without predictive value on the evolution of the disease.

Performing spirometry is very important to identify a small but significant group of patients (10%) with low FEV1 values and without obvious symptoms and to guide treatment decisions that reduce asthma morbidity and mortality .

The higher rate of exacerbations was correlated with MMEF values of <80%, which gives this parameter a predictive value for the risk of disease progression.

Keywords: bronchial asthma, spirometry, monitoring

Neuroendocrine tumours

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Co-authors: Denisa-Maria Borla[1], Răzvan-Andrei Gligor[1], Mădălina-Maria Negrea[1]
Coordinator: Dr. Alexandra-Elena Borla[1]

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Background
Neuroendocrine tumours are a heterogeneous group of malignancies that develop from neuroendocrine cells found in different organs and tissues. These tumours can be slow or fast growing and their clinical and biological characteristics can vary considerably.

Objectives
The objectives of the paper are to evaluate the efficacy of surgical excision in the treatment of neuroendocrine tumors, especially in terms of survival and control of associated symptoms, and to analyze paraclinical laboratory results such as APTT, PT, bilirubin, creatinine, glycaemia, liver enzymes, serum amylase, plasma protein leveles, creatinphosphokinase, LDH, serum sodium, serum potassium, serum haemoglobin, leukocytes and neutrophils to assess relevant biomarkers in the management of neuroendocrine tumours.

Materials and methods
Twenty patients with confirmed diagnosis of neuroendocrine tumour were followed up over a 5-year period (2017-2021). Of these, just over half were female (56.62%). The sex ratio was 1.3:1 female:male. Statistical analysis was performed using Microsoft Office Excel and IBM SPSS Statistic 28.0 software (IBM Corporation, USA, 2022). Descriptive statistics were calculated for all variables.

Results
The average age in the group was 60 years, with a minimum age of 20 years and a maximum age of 81 years. The mean age among male patients was 55 years, while in the group of female patients it was 66 years. This difference did not reach the threshold of statistical significance. All deceased patients were female. Analysis of paraclinical parameters showed significant differences between the deceased and cured patients. APTT and prothrombin time were significantly higher in deceased patients, suggesting an association between coagulation dysfunction and adverse outcome of neuroendocrine tumours. Direct and total bilirubin, serum creatinine and serum urea levels were significantly higher in deceased patients compared to cured or improved patients. These differences suggest the involvement of the liver and renal system in the adverse outcome of neuroendocrine tumours. The majority of patients had primary tumors in the intestine (69.57%), followed by pancreas (26.09%) and lungs (4.34%). Distant metastases were seen in 61% of patients, with liver and lung metastases being the most common.

Conclusions

Neuroendocrine tumors affect both men and women. Patients of any age may be involved. Patients with neuroendocrine tumours are at significant risk of complications and death. The prognosis is more reserved among women. Liver and lung metastases are most common. Surgery is the main treatment, but drug therapy and radiotherapy can be used as adjuvant.

Keywords: management, surgical, neuroendocrine, surgical, radiotherapy

Whole Exome Sequencing's role in the diagnostic odyssey of neurodevelopmental disorders

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Background
Elevating our grasp of medical genetics, Whole Exome Sequencing (WES) is taking us a step forward to personalised medicine. Whereas gene panels look for specific gene's variants, WES has a more global view over the patient's DNA, offering an unbiased approach in identifying rare genetic disorders. Neurodevelopmental disorders (NDDs) are a group of conditions that affect cognitive, motor and social functions. Because of the genetic heterogeneity, the clinical diagnosis challenge and the importance of early intervention that NDDs impose, it's obvious how WES could shorten the diagnostic odyssey in the best interest of the patients.

Objectives
This research's objective was to see how WES testing can be used to look for the underlying etiology of NDDs in order to provide the best medical care for the patients.

Material and method
We aimed to test patients with NDDs over a period of three months at the Medical Genetic Outpatient Clinic Regina Maria Cluj-Napoca. There were a total of 21 patients who presented with NDD-like phenotype and underwent WES testing.

Results
The results showed either pathogenic variants or variants of uncertain significance (VUS) for 6 patients.
Remarkably, one patient marked the first diagnosed case of ODLURO syndrome in Romania. The patient inherited a variant of the KMT2E gene from his father, who also presents autistic-like behaviour. The mutation, in this case, being very relevant for the parents' desire to have more children.
Genetic testing showed another patient's hemizygosity for a chromosome X duplication, indicative of the pathogenic MECP2 duplication syndrome. The last four patients exhibited modifications in their exomes that were also linked to NDD symptoms.
All patients presented mostly the same symptoms, but each one of them had a distinct gene defect. Thus, it's understandable that WES is the perfect tool to look for the etiology as there would be hundreds of possible genetic targets. There's more room for progress as WES testing didn't manage to unravel the etiology for 15 patients, showing there could be more epigenetic or non-genetic causes for the NDDs.

Conclusions
To sum up, our research showcased the effectiveness of WES in identifying distinct gene defects associated with NDDs. Notably, WES revealed novel diagnoses, such as ODLURO syndrome and MECP2 duplication syndrome, emphasizing its role in expanding our understanding of genetic disorders. Overall, WES is a new genetic technology that is paving the way for more targeted and individualised medical care for patients with neurodevelopmental disorders.

Keywords: Whole Exome Sequencing, neurodevelopmental disorders, ODLURO syndrome, MECP2 duplication syndrome, genetic testing

The Obesity-Menopause Conundrum in Breast Cancer – The Risk of Weight and Menopausal Status in Cancer-treated Patients

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Background
Breast cancer (BC) represents the most common malignancy in women, with incidence on the rise. While a polymorphic disease, obesity is one of the factors incriminated to increasing the risk of developing BC and to a worsening prognosis. Menopausal status is the fulcrum on which the association between obesity and BC reputedly hinges. One of the most consistent associations has been its increased risk for post-menopausal BC in contrast to a null or inverse association in pre-menopausal women. Obese post-menopausal women have an increased risk of hormone receptor-positive BC, whereas pre-menopausal women have a higher proportion of triple-negative BC, risk of recurrence, and mortality rates reported.

Objectives
The study aims to assess different clinical, imagistic, and biological determinants of fat mass and gonadotropic axis in BC-treated patients according to their menopausal status at diagnosis.

Material and methods
Seventy-eight BC female patients evaluated in the Endocrinology Department for metabolic or endocrine disease associated with BC treatment were included in the current cross-sectional analysis. All women were at menopause, either physiologic or therapeutically induced. Fat mass was assessed by dual-energy X-ray absorptiometry (DEXA), with data on total fat mass, subtotal fat mass (without head region), and trunk fat mass measured in grams (g) or as percentage (%) of total body mass. Immunoassays were used to determine serum hormone levels of insulin, FSH, estradiol, estrone, and total testosterone. Insulin resistance was calculated by the HOMA-IR index.

Results
Post-menopausal diagnosed women showed statistically significant higher BMI ($t=-3.159$, $p=0.002$), abdominal ($t=-2.305$, $p=0.036$), and hip circumference ($t=-2.683$, $p=0.009$), but not hip/abdominal ratio ($t=-0.647$, $p=0.520$); also, they had significantly higher body fat mass (either total, subtotal, or trunk fat; $p<0.05$). No statistically significant differences were observed in fasting glucose ($t=-0.800$, $p=0.426$), cholesterol ($t=-0.566$, $p=0.573$) or triglycerides ($t=0.969$, $p=0.336$). Insulin levels, however, were significantly increased in the post-menopausal group ($t=-2.420$, $p=0.018$), and subsequently, the HOMA-IR index was significantly increased ($t=-2.305$, $p=0.024$). Regarding sex hormones, no statistically significant differences were observed in circulating estrogens (estradiol and estrone) nor androgens (total testosterone and calculated free androgen index). However, for both pre-menopausal and post-menopausal groups, no statistically significant differences were observed in sex hormones according to weight status (obese vs non-obese).

Conclusions
This study shows a significant difference between weight status and risk of BC regarding menopausal status, supporting obesity as a risk factor in post-menopausal patients and a protective factor pre-menopausal, underlying the polymorphism of breast cancer disease.

Keywords: breast cancer; obesity; menopause.

1.4. Clinical Surgical

The surgical treatment of superior thoracic outlet syndrome

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Background
Thoracic outlet syndrome (TOS) was first described by Peet, in 1956, and encompasses three interrelated syndromes characterized by the compression of nerves, arteries or veins in the upper chest and lower neck region. The current treatment protocols involve a combination of conservative and surgical approaches, tailored to the type of TOS and the severity of symptoms.

Objectives
The primary objective is to promote the first rib resection as the most suitable therapeutic option for the above-mentioned pathology that is refractory to the conservative treatment. Secondary, the paper will enlighten the impact of surgical treatment on long-term outcomes, including pain relief, functional improvement, and patient satisfaction, following surgical treatment for TOS.

Material and Method
The present paper summaries and analyses the data from 318 articles published in English, in free full text listed on PubMed databases, in the last 5 years. Prospective cohort studies of interest and comparative studies that meet the inclusion criteria were encompassed. Furthermore, the paper illustrates our surgical technique based on case reports.

Results
In terms of surgical management, a standardized surgical approach has not yet been established. However, the largest series published in the literature indicates a high success rate with transaxillary first rib resection. The rationale of this approach is that the first rib forms the common denominator for all causes of nerve and vascular compression in this region, so that its removal generally improves symptoms Nonetheless, serious complications such as scapular winging and snapping, as well as pneumothorax, have been linked to this procedure. Additionally, some studies have reported a high recurrence rate. Consequently, to mitigate the risk of complications and recurrence, the supraclavicular approach has been recommended for first rib resection and scalenectomy. Besides these two surgical approaches above-mentioned, the video-assisted thoracic surgery (VATS) approach is a superior alternative to the classic transaxillary access for treatment of TOS. The visualization of the entire length of the first rib and the neurovascular bundle is excellent, enabling thorough posterior resection. Potential complications linked to rib resection for TOS encompass chylothorax, paresthesia, neurovascular damage, and persistent pneumothorax.

Conclusions
The first rib resection, regardless of the indication, is an operation with very good results in well-selected cases and can be performed with very low morbidity and negligible mortality.

Keywords: thoracic outlet syndrome, first rib resection, transaxillary approach, supraclavicular approach, VATS approach

Management of mediastinal parathyroid adenoma: diagnostic and therapeutic approach

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Background
Parathyroid adenoma is a benign tumor of the parathyroid glands which accounts for the majority of cases of primary hyperparathyroidism (80-85%). Abnormal migration during embryogenesis can determine an ectopic placement of the parathyroid glands, encountered in up to 16% of cases. A frequent site that should not be overlooked is the mediastinal position, where the gland is primarily intrathymic.

Objectives
The primary objective is to present the optimal management of the pathology by analysing the methods of diagnosis and imaging, as well as the therapeutical approach. Secondary, the paper will enlighten the techniques with the greatest therapeutic advantage and the least adverse effects.

Material and Method
The scientific paper summaries and analyses the data from 15 out of 40 identified articles published in English listed on PubMed databases, in the last 20 years. Comparative studies, reviews, meta-analyses and case series that meet the inclusion criteria were included. In addition, the paper illustrates the elected surgical treatment and technique by means of a case report.

Results
An integral part of the treatment is preoperative localisation of the ectopic gland. Ultrasonography, while useful in the neck region, has been proven inefficient for detecting mediastinal parathyroid adenomas. Preoperative ^{99m}Tc-MIBI scan has been utilised for detection of ectopic glands, reporting a sensitivity between 80 and 90%. Computerized tomography (CT) and magnetic resonance imaging (MRI) can also contribute to the diagnosis, but used alone they possess lower sensitivity (65% and 75% respectively). Better detection can be achieved with single emission photon computer tomography (SPECT) in association with CT(SPECT/CT). Four-dimensional CT has proved to be useful when the aforementioned investigations have failed, but the dose of radiation is higher. Positron emission tomography (PET) radiotracers (^{11}C-methionine, choline) could become a better alternative, however it is still limited by the high cost and low availability. Currently, the first line of imaging is comprised of ^{99m}Tc-MIBI associated with SPECT/CT. The most effective therapeutical option remains the surgical removal of the abnormal intrathymic glands. The small number of cases have halted the establishment of a standard approach. Video-assisted thoracoscopic surgery has been shown to have less complications and a lower mean hospital

stay than a median sternotomy. A cervical incision approach can be used, but it has proven ineffective in lower mediastinum sites.

Conclusion
Mediastinal parathyroid adenoma is a rare and challenging pathology, successful management depending on preoperative localisation which enables the use of minimally invasive surgical treatment.

Keywords: mediastinal parathyroid adenoma, parathyroid imaging, hyperparathyroidism, minimally invasive thoracic surgery

Inguinal hernia management

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Background
Hernia is a condition in which an organ or tissue protrudes through a weakened area of the abdominal wall. Hernia management involves assessment, diagnosis and treatment. The collaboration of surgeons with anaesthetists and radiologists is very important.

Objectives
The main aim of this paper is to evaluate the differences between elective surgery and emergency surgery patients .Thus, demographic characteristics, type of surgery and paraclinical data are followed.

Material and method
50 patients with inguinal hernia are followed retrospectively over one year. Patients with incomplete data were excluded from the study. All information collected from patients was centralized in a database created using Microsoft Excel; IBM SPSS Statistic 29.0 (IBM Corporation, USA, 2023) was used for statistical analysis. Independent groups t-student test was used for data comparison. A p-value <0.05 was considered statistically significant.

Results
The group consisted of 50 patients of which 11% were female and 89% male.Of these, 50% had emergency and 50% elective interventions.The average age in the group was 60 years, with a minimum of 21 years and a maximum of 92. 37% of the patients were from urban areas and 63% from rural areas. A much higher percentage of rural patients underwent emergency surgery compared to urban patients.The average age of patients who underwent emergency surgery was significantly higher than those who underwent elective surgery. Only 1% of elective surgery patients and 5% of emergency surgery patients had postoperative hydrocele.1% of patients who underwent emergency surgery developed postoperative phlegmon, compared with electively operated patients in whom this complication did not occur.

Conclusions
Patients undergoing emergency surgery have a higher rate of postoperative complications such as intestinal obstruction, the phlegmon.The mesh was used more often among emergency surgery patients. The recurrence rate is as high among emergency patients as it is among elective patients.Elective procedures were associated with lower serum glucose values compared to emergency procedures.Emergency surgery patients had slightly higher ASA scores, indicating a more severe physical condition before surgery.

Keywords: hernia, emergency, male, hydrocele, phlegmon

The Management of Acute Mesenteric Ischaemia

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Background
Acute mesenteric infarction is caused by a sudden drop in blood flow. Nonspecific symptomatology together with the fast progression contributes to a high mortality rate of about 60-80%.

Objectives
The aim of the study is to highlight the difficulty of establishing a diagnosis based on a non-specific clinical picture and the importance of risk factors and clinical variables. The study also aims to follow the evolution of patients and their condition at discharge.

Material and method
Fifty-eight patients with a diagnosis of mesenteric infarction with diagnosis code K55.0 are being followed from February 2018 to July 2022. Patients with incomplete data or those who died before surgery were excluded. Statistical analysis was performed using Microsoft Office Excel and IBM SPSS Statistic 29.0 (IBM Corporation, USA, 2022). Descriptive statistics were calculated for all variables. The $\chi 2$ test (Chi-Squared Test) with or without Yates correction was used for comparisons between categorical variables.

Results
The study shows a higher prevalence among men than women. The average age in the group was 70 years. In the female patient group, the age was higher compared to the male patient group. 81% of the patients in the group were pensioners, of whom 64% showed improvement at discharge. 29% of the patients in the group died after surgery. 59% of patients had diabetes, 55% hypertension, 84% were obese, 62% had dyslipidemia. 81% of patients were non-smokers. In the batch, the average number of secondary diagnoses was 11, with a maximum of 23. Renal function was assessed using creatinine and urea. The mean creatinine value was 2 mg/dl. Sindromul inflamator asociat injuriei vasculare a fost evaluat folosind valoarea numarului de leucocite si neutrofile circulante. Inflammatory syndrome associated with vascular injury was assessed using circulating leukocyte and neutrophil counts.

Conclusion
Although smoking is an independent risk factor in the pathogenesis of vascular disease, 81% of the patients in the group were non-smokers. This demonstrates that the presence of other comorbidities such as diabetes mellitus, hypertension, dyslipidaemia and ischaemic heart disease may have a more important role in the pathogenesis of the disease.

Keywords: ischaemia, sudden, elderly, comorbidities, nonspecific

Innovation in the Treatment of Myelomeningocele: Advanced Approach to Correction Through Fetal Surgery

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Background
Fetal surgery, also known as intrauterine surgery, has evolved significantly over the years. This specialized branch of surgery focuses primarily on treating congenital conditions and abnormalities of the developing fetus. Prenatal repair of myelomeningocele (MMC), the most common and severe form of spina bifida, is an exceptionally delicate surgical procedure in which fetal surgeons open uterus and close the opening behind the baby while it is still in the womb. Because spinal cord injury progresses throughout the gestational period, prenatal repair of myelomeningocele can prevent further damage.

Objectives
The aim of this study is to meticulously analyze the surgical approach to myelomeningocele and ascertain its outcomes. By delving into this intricate procedure, we aim to shed light on the efficacy and impact it holds in clinical practice, contributing to the advancement of neurosurgical care.

Materials and Methods
The research was conducted on PubMed using the keywords "fetal surgery," "spina bifida," "myelomeningocele," and "prenatal." Scientific papers were selected based on inclusion criteria, which involved patient groups treated with both prenatal and postnatal surgery, recovery period, postoperative well-being of patients, operating time and the effectiveness of the procedure itself. Articles mentioning patients with major complications were excluded. Risk of bias was not assessed, and PRISMA criteria were used for data synthesis.

Within the carefully curated selection of 12 articles, we have delved into two impactful studies that contribute significantly to the field. The initial study, led by the esteemed National Institute of Child Health and Human Development (NICHD), meticulously scrutinized a cohort of 183 patients treated between 2010 and 2020. Notably, 91 patients underwent prenatal surgery, while 92 opted for postnatal intervention.

The second study, conducted at the Saint Louis Fetal Care Institute, methodically investigated a cohort of 58 patients, all of whom received prenatal treatment, spanning the years 2011 to 2017. Thus, this scientific undertaking illuminates a collective insight derived from a comprehensive study group of 241 patients.

Results
According to the first study, children from the group that underwent prenatal surgical intervention showed a significantly higher likelihood of achieving independent mobility (44.8% compared to 23.9% in the group that underwent surgery after birth). Subsequent research indicates that prenatal repair of myelomeningocele improves motor outcomes at

the age of 30 months compared to postnatal repair. Within the cohort of 58 patients analyzed in the second study, 2 unfortunately succumbed to prematurity (3.44%), and 30 required treatment for hydrocephalus (51.72%).

Conclusion

Fetal surgery for spina bifida brings remarkable benefits, notably reducing the need to divert fluid from the brain, enhancing mobility, and increasing the likelihood of independent walking for the baby. This intervention addresses and mitigates the effects of spina bifida, a congenital condition impacting the spinal cord. By surgically repairing the myelomeningocele (MMC) prenatally, the procedure contributes to improved long-term outcomes for affected individuals, preventing ongoing damage and fostering a foundation for healthier development. This proactive approach underscores the transformative impact of medical innovation on the lives of those with spina bifida.

Keywords: Fetal surgery, intrauterine, myelomeningocele, spina bifida, independent walking, prematurity , hydrocephalus

Treatment of Biliary Obstruction- Endoscopic Retrograde Cholangiopancreatography vs Traditional Surgery

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Background
Cholecystolithiasis with obstruction of the bile duct is a common medical issue. Endoscopic retrograde cholangiopancreatography (ERCP) and laparotomy with choledochotomy are commonly used in treating this condition. ERCP is a minimally invasive procedure allowing for the assessment and treatment of biliary tract problems, whereas laparotomy with choledochotomy is the traditional method involving a large incision in the abdomen.

Objectives
This review aims to compare these two surgical methods in terms of: hospitalization period, recovery period, operating time and postoperative well-being.

Materials and Methods
The research was conducted on PubMed using the keywords "gallstone," "ERCP," "choledochotomy," and "stone extraction." Scientific papers were selected based on inclusion criteria, which involved patient groups treated with both ERCP and laparotomy, hospitalization period, recovery period, postoperative well-being of patients, operating time and procedure effectiveness. Articles mentioning patients with major complications and individuals over the age of 70 were excluded. Risk of bias was not assessed, and PRISMA criteria were used for data synthesis.

Results
Out of the total of 18 articles found, 10 studies were selected. The cohort comprised 1423 patients, with 712 treated using ERCP and 712 through traditional surgery. The patient group treated with ERCP had a hospitalization period of 1-2 days, with postoperative pain that was easier to manage and an operating time of approximately 15 minutes. Meanwhile, the group treated with the traditional method experienced moderate to intense pain, and the hospitalization period ranged from 7 to 20 days and with an operating time of approximately 90 minutes. Minimally invasive surgery patients had a recovery period of up to 3 weeks compared to the other group, where the maximum recovery period was 7 weeks.

Conclusion
ERCP represents a less invasive alternative to laparotomy with choledochotomy in the treatment of gallstone disease. Understanding the advantages offered by ERCP, such as rapid recovery, reduced complications, and diagnostic precision, makes this method an attractive option in the management of gallstones. These advantages also encourage further research into these treatment methods.

Keywords: Common bile duct, gallstones, laparotomy, complications

Section 2

Poster Presentations

Clinical Medical and Basic Medical Science

2.1.Winners of the competition

First place

Is our destiny written in our genes? – a hypertrophic cardiomyopathy case

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Introduction
Hypertrophic cardiomyopathy (HCM) is the most frequently inherited heart disease and an important cause of sudden cardiac death in young patients, who are often unaware of their underlying condition. Mutations in MYBPC3 and MYH7 are the most frequent genetic cause of HCM and are attributed to more than 50% of all HCM cases. The clinical heterogeneity of HCM requires ways to identify patients at a high risk of complications and to evaluate their quality of life.

Case Report
A three-year-old male patient was admitted to the hospital due to Rx abnormalities (cardiomegaly) detected during examination for an episode of pneumonia. Paraclinical examinations (ECG, echocardiography, cardiac MRI) led to the diagnosis of hypertrophic cardiomyopathy. The genetic investigation (NGS) was performed and revealed two pathogenic variants, c.1224-52G>A (Intronic) and c.3124del (p.Thr1042Leufs*4), identified in MYBPC3.
MYBPC3 gene is associated with a spectrum of autosomal dominant cardiomyopathy conditions. Furthermore, 5 additional variants of uncertain significance were also identified. The family genetic testing revealed that the patient's mother was carrying the pathogenic variant, c.1224-52G>A (Intronic), identified in MYBPC3 and that the patient's father was carrying the pathogenic variant, c.3124del (p.Thr1042Leufs*4), identified in MYBPC3. Prior to the genetic testing, the parents showed no clinical signs of cardiac pathology.

Discussion
HCM is an archetypical single-gene disorder with an autosomal dominant pattern of inheritance. Therefore, a single mutation is usually sufficient to cause the disease, however, with variable penetrance and expression.
The variability of the phenotype is due, at least in part, to the causal mutation acting in concert with several other genetic and nongenetic influences. Approximately 60% of patients with HCM have a recognisable familial disease.

Conclusion
Although specific genotype-based treatments for HCM are not yet available, positive genetic test results confirm the aetiology of the disease and enable mutation-specific confirmatory testing of the appropriate family members.

Keywords: MYBPC3, cardiomyopathy, hypertrophic

Second Place

Multisystemic Inflammatory Syndrome in Children: an emerging cause of coronary artery aneurysms

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Introduction
Initially, children infected with SARS-COV-2 were regarded to be either asymptomatic or presenting mild symptoms. However, in light of recent findings, a new entity was described as multisystemic inflammatory syndrome in children (MIS-C), associated with SARS-COV-2, consisting of a severe hyperinflammation state with a wide array of symptoms. Due to the vital prognosis, we will be discussing the cardiovascular involvement.

Case report
We report the case of a 7-year-old boy referred with a persistent fever for the past month. His recent history included cervical lymphadenopathy, hepatosplenomegaly, and myalgia. Upon admission, the physical exam also indicated bilateral conjunctivitis and strawberry tongue. Further laboratory investigations showed high levels of inflammatory markers including CRP, ESR, and IL-6 as well as positive SARS-COV-2 Ig G antibodies. A giant right coronary artery aneurysm measuring 6/9mm (Boston Z score of 10) was found on cardiac ultrasonography. Thus, the patient fulfilled the WHO criteria for SARS-CoV-2-associated multisystem inflammatory syndrome. He was treated with intravenous immunoglobulins and methylprednisolone, as well as aspirin and Enoxaparine due to the possible thrombotic complications. Although both clinical and biological findings normalized, the coronary aneurysm showed no improvement at the 1,3 and 6-month follow-up.

Discussions
MIS-C is hypothesized to be caused by a post-infectious immune dysregulation which explains the improvement seen after immunomodulatory agents. Cardiac involvement is a frequent feature and consists mainly of left ventricular dysfunction, coronary artery aneurysms, and myocarditis. Given the overlapping features with Kawasaki disease, the leading cause of acquired heart disease in children, prompt diagnosis and treatment are essential for MIS-C patients.

Conclusions
Coronary artery aneurysms are reported in 6% to 24% of patients with MIS-C. Due to its novelty, the long-term evolution of cardiovascular sequelae remains unknown. However, given the possibility of severe cardiac ischemia at a young age, clinicians should be familiar with this entity and ensure a long-term follow-up.

Keywords: MIS-C, coronary artery aneurysm, SARS-COV-2, children

Third Place

A medical association: Autism spectrum disorder caused by an oncologic predisposition syndrome

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Introduction
Cowden syndrome is a rare autosomal dominant disease characterized by the development of multi-systemic hamartomas and increased risks of breast, thyroid, renal and endometrial cancers. Most patients with Cowden syndrome present mutations of the phosphatase and tensin homolog (PTEN) gene on chromosome 10, being one of the PTEN hamartoma tumor syndromes (PHTS). The involvement of this gene in the pathogenesis results in the variability of symptoms of this disease. An unusual manifestation is the prevalence of cognitive disorders and autism spectrum disorder (ASD).

Case report
We report the case of a 10-year-old male patient with a family history specific for PHTS (uncle with macrocephaly), currently diagnosed with right thyroid nodule (under evaluation and treatment). The patient presents characteristic symptoms of Cowden syndrome: growth retardation, premature puberty, macrocephaly and hyperpigmentation of the glans. Furthermore, the patient is known with various other types of tumors, such as axillary hamartomas and right testicular lipomas (previously excised). As far as cognitive disorders are concerned, the patient developed ASD symptoms starting from the age of 1 year and 6 months, with the language function being the most affected. Considering the particular phenotype, an NGS-21 gene panel was recommended. The molecular analysis identified the heterozygous c. 414T>G, p(Tyr138*) pathogenic variant in the PTEN gene.

Discussions
The variability of the Cowden syndrome’s symptoms makes this disease hard to diagnose. Furthermore, it is one of the few causes of non-idiopathic ASD.

Conclusions
The fact that the PTEN gene is also involved in brain development leads to the unique association between two apparently different conditions: an oncologic predisposition syndrome and ASD.

Keywords: ASD, Cowden Syndrome, PHTS

First Honorable Mention

Rituximab-Induced Kaposi Sarcoma in a HIV-Negative Patient with Large B-Cell Non-Hodgkin Lymphoma

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Introduction
Diffuse large B-cell lymphoma is the most common form of non-Hodgkin`s lymphoma. The standard treatment regimen is R-CHOP (rituximab, cyclophosphamide, doxorubicin, vincristine, prednisolone). Rituximab is thought to exacerbate Kaposi sarcoma, a disease mostly encountered in immunocompromised HIV-positive patients, the aetiologic agent being KSHV/HHV8.

Case report
We describe the case of a 77-year-old Caucasian male who presented in the clinic with a left cervical mass and upon HP/IHC evaluation was diagnosed with stage IIA Large B-cell NHL. The blood tests were normal and a CT scan revealed laterocervical adenopathies. After surgical removal of the mass, from May to October 2021, he underwent eight 3-week cycles of R-CHOP, which were well tolerated. Another CT scan was made in November which showed no suspicious lesions. After the 6^{th} cycle the patient presented with purplish spots on the back of the hands, which exacerbated in December 2021, spreading to the upper arm and the thigh. The histopathological appearance was compatible with the diagnosis of Kaposi sarcoma; the IHC staining was positive for HHV8, but the HHV8 and HIV serological tests came out as negative. From February to July 2022 he underwent 12 cycles of paclitaxel, having a favourable evolution.

Discussions
What makes this case stand out is that our patient did not have a history of KS. Rituximab may be involved in latent HHV8 reactivation due to B-cell depletion, increasing the chances of KS development in HIV-negative patients with a compromised immune system due to another cancer. Such events are rare, limited to a few case reports in the literature and the underlying mechanism has not been yet elucidated, requiring further studies.

Conclusions
Clinicians should be vigilant when administering rituximab alongside other immunosuppressive therapies in cancer patients and should resort to HHV8 screening in those at risk of infection.

Keywords: Kaposi Sarcoma, Lymphoma, Rituximab, HIV

Second Honorable Mention

An unusual Burkitt lymphoma case with particular pathology and unexpected evolution

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Introduction
Burkitt lymphoma is a rare aggressive disease, more commonly found in children, but with worse prognosis in adults. Two clinical variants were reported: sporadic (unknown etiology) and endemic (immunodeficiencies). Our case report highlights a sporadic primary gastric Burkitt lymphoma, a non-Hodgkin subtype, with only a small number of cases reported worldwide.

Case Report
Disease onset occurred in June 2023 when a 10-year-old boy presented fever initially treated as an upper respiratory tract infection by the primary physician. The symptoms evolved with jaundice and he was suspected of hepatitis. During conservatory treatment for hepatitis, massive coffee-ground vomiting appeared. CT-exam revealed a thickening of the gastric wall with bile duct compression explaining the initial symptoms. The patient was admitted into the pediatric oncology department where we opted for transcutaneous eco-guided biopsy resulting in diagnosing the patient with Burkitt lymphoma. The staging was completed with bone marrow aspiration and a spinal tap, ranking it stage III. He underwent chemotherapy with Rituximab based protocol showing quick response confirmed by the CT scan. The side effects reported were pancytopenia with fever in 2 cycles, digestive toxicities (nausea, vomiting, mucositis) and hepatic cytolysis.

Discussions
Burkitt lymphoma is one of the least common forms of gastric cancer affecting children. This tumor should be treated aggressively, chemotherapy showing increased sensitivity. Long-term survival rates are reported, whereas surgery has a small role. Transcutaneous biopsy proved to be more accessible in our case than endoscopy, due to the extension of the tumor in the superficial hypogastric region. The initial symptoms mimic different conditions and careful considerations should be given to establish the diagnosis and start treatment immediately. Delayed diagnosis and treatment lead to death due to this highly curable disease.

Conclusions
We reported the case of a rare lymphoma with an even rare presentation. Quick diagnosis and aggressive treatment lead to complete response, which is confirmed at the monthly follow-ups.

Keywords: Burkitt lymphoma, Rituximab, transcutaneous biopsy, hepatitis

2.2. Basic Medical Science

Histamine intolerance caused by genetic DAO deficiency

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Introduction

Histamine intolerance (HIT) defines the intolerance of histamine manifested at the ingestion of food. It is caused by a deficiency of the histamine-degrading enzyme diamine oxidase (DAO) or an existing imbalance between histamine and DAO. The polymorphisms of the DAO or HNMT, drugs and foods that inhibit the activity of these enzymes, foods rich in histamine or promote histamine release, respectively dysbiosis are the main etiological factors of HIT. This condition occurs in 1% of the population, females over the age of 40 are predisposed to develop HIT, possibly linked with the reduction of sexual hormones.

Case report

We report the case of a 29-year-old woman presenting the following symptoms: paroxysmal episodes suggestive for HIT: migraines, vertigo, fatigue, nasal congestion, rhinorrhea, dyspnea, dermatographic urticaria, irritable bowel syndrome, tachycardia, anxiety. Following genetic testing, the results revealed a heterozygous DAO status for the variants rs2052129 GT, rs10156191 CT (associated with higher risk of migraines and migraine susceptibility), rs2268999 AT, and WT (wild type) for rs1049742 WT.

Discussions

Considering the predisposing genotype of the DAO enzyme in the absence of other DAO inhibiting conditions, the diagnosis of histamine intolerance secondary to deficient DAO activity was put. Though literature does not specify severe forms of histamine intolerance among heterozygous carriers; the manifestations imply a multifactorial etiology, therefore, a multidisciplinary approach is needed with the implementation of a specific DAO diet and intestinal microbiome restauration.

Conclusions

1. Histamine intolerance represents the imbalance between the accumulated histamine in the body and the capacity to degrade it.
2. Some individuals are genetically predisposed towards histamine intolerance presenting individual variability in the DAO enzyme expression and identifying polymorphisms associated with gastrointestinal pathology.
3. Therapeutic objectives include nutritional counseling, DAO substitution; DAO food supplements; copper, zinc, vitamin B6 supplements and avoiding DAO inhibiting medication.

Keywords: Histamine, Intolerance, DAO, polymorphism

A study of BRCA mutations in prostate cancer

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Introduction
Prostate cancer is the second most frequent neoplasia in men and the second leading cause of death in developed countries. Understanding its molecular profile may contribute to a more accurate diagnosis, to a better prognosis definition and therapeutic choice. BRCA1/2 mutations are reported to increase the aggressiveness of the disease.

Objective
Molecular diagnosis of prostate cancer by assessing BRCA1/2 mutations and their relation with the aggressiveness and the stage of the disease.

Material and Method
111 patients with prostate cancer were enrolled. The diagnosis was confirmed by PSA serum level, biopsy, and imaging. All the patients signed the informed consent. We recorded the Gleason score, the stage of the disease and the initial PSA value. A blood test for BRCA evaluation was performed to all the patients upon enrollment.

Results
Most of the patients (83,8%) were diagnosed with advanced prostate cancer (stage IV). The BRCA2 mutation was detected in 5,4% of the patients, while the BRCA1 mutation was not present in any of the patients. 1,8% of the patients presented the ATM mutations. There were no statistically significant differences between patients with BRCA 2 present and those without it concerning the clinical characteristics and laboratory parameters. Although Gleason score was higher in patients with BRCA 2 present, the statistical significance threshold was not reached.

Conclusion
The frequency of BRCA mutations in the studied population is in accordance with the data reported in the literature. We have not found any statistically significant correlations between the BRCA mutations and the initial stage of the disease, the Gleason score and the initial PSA value.

Keywords: Prostate adenocarcinoma, BRCA1/2 mutations, Disease stage, Gleason score, Initial PSA value

Lynch associated breast cancer diagnosed by NGS germline panel

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Introduction
Lynch Syndrome is a rare, autosomal dominant hereditary cancer syndrome, caused by mutations in the MMR (DNA mismatch repair) genes MLH1, MSH2, MSH6, PMS2 or EPCAM. Patients with Lynch Syndrome present high microsatellite instability, MSI-H, having cancer predisposing phenotype.

Case presentation
We report the case of a 49-year old woman, premenopausal, with a family history positive for endometrial cancer. The patient's oncological history began in May 2023, when a mammography, followed by an ultrasound revealed minimal architecture distortion in the right breast. The histopathology exam confirmed a Luminal B invasive breast carcinoma.

Discussions
Given the discovery of the tumour at an early stage, along with the particularity of three family members being diagnosed with endometrial cancer, genetic testing was the next step taken. The Oncotype Dx multigene test showed an RS (recurrence score) of 16, chemotherapy presenting lower than 1% benefits for this patient. Following the NGS-based germline panel test, a heterozygous mutation in MSH6 was discovered, which led to the final diagnosis of Lynch Syndrome.
The patient underwent hormone therapy (with Tamoxifen combined with Goserelin) for 3 months, followed by Breast conserving surgery and SNLB. Adjuvant treatment included External Beam Radiation Therapy and Hormone Therapy.

Conclusions
This case is unique due to the convergence of two significant factors: the discovery of MSH6 mutation in a patient with breast cancer and the subsequent diagnosis of Lynch Syndrome. The rich family history of endometrial cancer emphasises the importance of genetic testing in uncovering hereditary cancer syndromes, shedding light on the relationship between breast cancer and Lynch Syndrome.

Keywords: Lynch Syndrome, Breast Cancer, MSH6 gene, Hormone Therapy.

2.3. Clinical Medical

Aseptic meningitis: a rare complication of rheumatoid arthritis

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Introduction
We will be presenting a rare case of rheumatoid meningitis, an infrequent complication that can occur in patients with rheumatoid arthritis (RA), an autoimmune and inflammatory disease which mostly targets the joints. Rheumatoid meningitis can manifest with cognitive impairment, cranial nerve palsy, severe headaches, stroke-like symptoms and seizures. Neuroimaging, especially FLAIR MRI shows hypersignals along the meninges, layers which cover and protect the brain and the spinal cord.

Case description
A 61-year-old woman, with a long history of RA, under treatment with Leflunomide for the past 7 years, was admitted to the Neurology Clinic for gait disturbances, coordination impairment on the left side, dysarthria, and involuntary movements with focal onset in the left lower limb. Hypersignals were noticeable on the FLAIR MRI sequence, especially in the cortex of the right hemisphere. Laboratory investigations showed that the rheumatoid arthritis was kept under control with the treatment (RF was 12.07, normal value<14 IU/ml). CSF analysis showed normal glucose levels, elevated protein levels, and the presence of white blood cells, especially mononuclear ones (30 cells/mm^3). Moreover, no malignant, viral, bacterial or fungal causes were identified through CSF analysis, proving that the meningitis was aseptic.

Treatment consisted of methylprednisolone pulses, 1g/day for 3 days. Leflunomide was changed to Azathioprine, 150 mg/day. Additionally, Levetiracetam, 1000mg/day, was administered to prevent other myoclonic seizures.

Discussions
Rheumatoid meningitis is a rare complication of RA, since between 2003 and 2018, studies reported under 50 cases, 59% of them women.

Conclusions
Differential diagnosis should be made through blood tests, CSF analysis and MRI. Treatment is usually with corticosteroids, such as methylprednisolone, to counteract the inflammatory effects of RA.

Keywords: Aseptic meningitis, Rheumatoid arthritis, Myoclonic seizures, CSF analysis, FLAIR MRI.

A case of Wilson's disease with neurological debut

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Introduction
Wilson's disease is a rare autosomal recessive disorder, in which unexcreted copper accumulates in tissues, resulting in hepatic, neurological and psychiatric symptomatology.

Case report
A 26-year-old male patient complaining of resting tremor affecting his hands and head was refered for a gastroenterology consultation after a cerebral MRI showed modifications suggestive of Wilson's disease. Clinical examination revealed a Kayser-Fleischer ring. On abdominal ultrasound, the liver appeared micronodular, with an irregular border, with signs of portal venous hypertension (splenomegaly of 18cm and hepatofugal portal flow). The upper endoscopy uncovered esophageal varices of grades 1 and 2. Based on these findings, a diagnosis of liver cirrhosis class Child Pugh A (5p) could be formulated.

A transjugular liver biopsy was performed and the histopathology was suggestive of liver cirrhosis and positive for copper stain (Rhodanine stain). Laboratory findings showed a high increase in urinary copper (579.6μg/24h) and low serum ceruloplasmin (0.071g/l). Genetic testing was negative for the H109Q mutation in the ATP7B gene.

The final diagnosis, based on a Leipzig score of 9 (certain diagnosis) was Wilson's disease. He currently follows treatment with zinc and Penicillamine (the dose was increased gradually from 500mg to 1250mg/day based on the levels of urinary Cu) with significant amelioration of both neurological signs and liver disease (the portal vein flow was gradually reversed and is now hepatopetal).

Discussions
Although negative for the H1069Q mutation, Wilson's disease is associated with a high number of pathologic mutations, so in this case, a negative genetic test could not reject the final diagnosis considering the specific clinical manifestations. Another particular aspect of this case was the portal venous flow, which reversed back to normal after treatment.

Conclusions
Although rare, Wilson's disease is a cause of chronic liver disease to be considered in coexisting hepatic and neurologic symptomatology.

Keywords: Wilson disease; tremor; liver cirrhosis; copper

CD8+ Mycosis fungoides: a rare subtype of the classic variant

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Introduction
Mycosis fungoides (MF) is the most common type of primary cutaneous T-cell lymphoma. The diagnosis may pose a challenge in the early stages and in atypical clinical presentations as it may simulate a large variety of common dermatologic disorders.

Case report
We present a case involving a 60-year-old male who was referred to the Hematology Department at the Regional Institute of Oncology (RIO) Iasi due to a 15-year history of psoriasis that was refractory to treatment. The patient exhibited an erythematous, scaly, ulcerous, and pruritic rash covering a significant portion of his body, prompting suspicion of T-cell skin lymphoma. Multiple skin biopsies supported the diagnosis of atypical MF. with CD8+, CD30+, Ki67 30% markers. Immunophenotyping from blood and bone marrow aspiration indicated the absence of abnormal T lymphocytes. In contrast, computed tomography (CT) imaging revealed multiple lymph nodes varying in size from 1.5 to 2 cm, which were too profound for sampling. The patient was refractory to initial treatment which included oral Methotrexate and second line therapy was initiated with Brentuximab, to which the patient showed good tolerance and favorable evolution.

Discussions
The diagnosis of MF is often challenging, due to the atypical aspect in early stages. The skin lesions simulated psoriasis and more thorough investigations were necessary for a correct interpretation. Moreover, CD8+ MF is a much rarer entity than its CD4+ counterpart, making the diagnosis more difficult for the pathologist.

Conclusions
This case report highlights the recurrence in misdiagnosis of MF. This is because the spectrum of clinical and histological features are seen in both benign inflammatory diseases and MF and more specific tests are necessary to reach an accurate diagnosis, including TCR clonality molecular studies.

Keywords: Mycosis fungoides, Psoriasis, Skin lymphoma, Rash

Elusive Gastrointestinal Bleeding: A Case Report of Complicated Jejunal Diverticulitis

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Introduction

A patient with lower gastrointestinal bleeding may present with a wide range of symptoms, from a display of light anemia to sheer hemorrhagic shock. Even though complicated diverticulosis is a known differential diagnosis in such cases, it is rarely uncovered while examining the small intestine, considering that more than 95% of diverticula are found in the colon.

Case Report

A 59-year-old female patient with a history of acute pancreatitis presents to the ER with hematochezia, blood clots in feces, diffuse abdominal pain, syncope and vomiting. The symptoms debuted two days prior. The digital rectal exam describes traces of fresh blood. Laboratory studies revealed an inflammatory syndrome and moderated anemia. CT angiography showed signs of enteritis.

The patient was scheduled for a colonoscopy. Due to the patient still experiencing bloody stools the procedure was visually limited, with no signs of hemorrhagic lesions on the colic wall. A polypectomy was performed for a sessile sigmoid polyp.

Following three successive episodes of hematochezia the patient is scheduled for a second angiography. The source of the bleeding is revealed to be a diverticulum located in the proximal jejunum.

Emergency exploratory laparotomy is performed, showing intestinal loops being dilated due to endoluminal bleeding. Five mesostenic diverticula are found The resection is followed by a jejuno-jejunal anastomosis. Histopathological examination confirmes the diagnosis of jejunal diverticulitis.

Discussions

Diverticulosis is predominantly found in the colon and is usually asymptomatic. Nevertheless, the small intestine is mostly inaccessible by means of endoscopy, which can lead to difficulty in locating these sources of hemorrhage. In such cases, CT angiography is advised.

Conclusions

Jejunal diverticulosis is an uncommon cause of lower GI bleeding. Most conventional imaging methods prove to be ineffective when confronted with this clinical entity. A quick diagnosis is crucial in preventing further complications.

Keywords: Jejunal, diverticulum, diverticulosis, bleeding, uncommon

A game of hide and seek: incidental cancer diagnosis using NIPT

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Introduction
Non-invasive prenatal testing (NIPT) is a screening method based sequencing cell-free DNA(cfDNA),found in placenta and maternal circulation. Boasting a remarkable 99% accuracy, with false positive and false negative results below 0.1% of cases, the purpose of this case report is to underline the causes of this enigmatic realm of inconclusive NIPT results, unraveling the complexities that render such outcomes unpredictable.

Case report
We present a case of a 40 years old woman, 18 weeks pregnant, who sought further investigations due to an inconclusive NIPT result. She had one previous pregancy that ended with a miscarriage after in vitro fertilization. An ultrasound by the obstretician revealed no signs of chromosomal or malformative abnormalities. He recommended NIPT screening, showing inconclusive results, and PrenatalSafe Karyo Plus, also with inconclusive results. NIPT identified several severe anomalies in chromosomes 3, 11, 12, 13, and minor anomalies in other chromosomes, without being able to precisely specify their cytogenetic/molecular characteristics.The analysis was repeated in another laboratory, but this investigator also provided indeterminant, so an amniocentesis followed by QF-PCR and chromosomal array analysis was conducted. After birth, confirmation of acute myeloid leukemia emerged.

Discussions
Contrary to expectations, a fetus with complex chromosomal defects exhibited normal ultrasound findings during gestation. Possible explanations include errors in methodology, placental mosaicism, the "vanishing twin" syndrome, or the presence of benign tumors and maternal malignancies.

Conclusions
Highlighting the complexity and difficulty of identifying hidden medical conditions during pregnancy, this case underscores the importance for a multidisciplinary approach and increase attention in managing maternal health. This is crucial to ensure the detection and proper treatment of any underlying conditions.

Keywords: prenatal screening method, maternal malignancies, inconclusive results, chromosomal abnormalities.

SIADH in a patient with advanced small cell lung carcinoma

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Introduction
Syndrome of inappropriate antidiuretic hormone secretion (SIADH) causes significant sodium depletion with plasma hypoosmolality, most frequently associated with small cell lung carcinoma (SCLC) where is characterized by ectopic secretion of vasopressin. We report a case of severe hyponatremia caused by SIADH and aim to emphasize the importance of early diagnosis and treatment for a better outcome.

Case Report
A 53-year-old male is transferred to the Intensive Care Unit (ICU) with history of SCLC stage IIIB under palliative radio and chemotherapy, hilar and mediastinal adenopathies, chronic obstructive pulmonary disorder stage III GOLD, bronchopneumonia, chronic smoking, rapidly progressive ataxia, sensory polyneuropathy, vestibulocerebellar syndrome and chronic hepatitis B.
The admission reasons are severe hyponatremia, hyperkalemia and acute kidney injury (AKI). The patient is conscious, cooperative, with symmetrical intermediate pupils, afebrile, with 14 points on Glascow Coma Scale. He is breathing on the face mask with O2, flow 12L/min, has bilateral disseminated rales, tracheo-bronchial load, SaO2 98%, stable, without vasoactive support, unmonitored diuresis, warm skin, without edema. Laboratory data revealed a sodium level of 104,8 mmol/L, potassium of 6,93 mmol/L, hypoalbuminemia, hypochloremia, elevated creatinine and urea levels, with a calculated filtration rate of 9 mmL/min.
An arterial line on the left radial artery, a central venous catheter and an urinary catheter were installed to monitor vital functions and the treatment was started with NaCl 5,85% 50mEq/50ml at 7 mL/h and furosemid.
In the second day is diagnosticated with SIADH, acute respiratory failure, protein-calorie malnutriton, immunodepression. Even with subtle improvments in sodium level, after three days in ICU, unfortunately the patient passed away.

Discussions
SIADH is found in 15% of patients with SCLC and can lead to neurological sequelae that can affect the long term outcomes.

Conclusions
An early diagnosis for paraneoplastic syndromes in malignacy patiens is challenging, but they need to be considered expecially in patient with SCLC.

Keywords: SIADH, small cell lung carcinoma, severe hyponatremia, paraneoplastic

The intriguing mimicry: Ovarian serous adenocarcinoma or Demon-Meigs Syndrome

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Introduction
Ovarian serous adenocarcinoma, constituting approximately 90% of all ovarian cancers, represents a subtype within epithelial cancer. Demon-Meigs Syndrome is characterized by a triad involving a benign ovarian tumor, ascites and pleural effusion, resolving after tumor resection.

Case report
This case report presents an 85-year-old patient with intermittent abdominal discomfort, intensifying over the past two months. The initial diagnosis, derived from a thorough physical examination, points towards ascitic, abdominal pain and anemic syndrome.
In terms of paraclinical investigation ultrasound imaging reveals the presence of ascites and multiple renal cysts. Elevated CA125 levels, an ovarian tumor marker, are evident in blood tests. Computed Tomography exposes multiple nodular lesions in the abdominal-pelvic region, alongside ovarian spaces exhibiting a heterogeneous structure. Pelvic Magnetic Resonance Imaging raises suspicion regarding malignancy in the left ovary. Further ascitic fluid samples undergo cytological scrutiny.
The patient was discharged with peritoneal carcinomatosis and suspicion of ovarian cancer, along with a recommendation for surgical intervention. The possibility of Meigs syndrome is considered in the differential diagnosis.
The patient returned after five days, exhibiting neurological complications. Cytoblock results, in corroboration with the presence of cerebral laconism confirmed by a CT scan, unveil a low-grade serous ovarian carcinoma in association with peritoneal carcinomatosis. Concurrently, a diagnosis of uterine leiomyoma surfaces, contributing to a pseudo-Meigs syndrome. The conclusive diagnosis encompasses ovarian serous carcinoma, peritoneal carcinomatosis, and leiomyoma, necessitating surgical consideration. After the anesthesiologist's consult, surgical intervention is deemed unadvisable. The patient is referred to an oncologist, for further monitoring.

Conclusion
In conclusion, this case highlights the importance of meticulous investigations in the context of differential diagnosis. Initially approached as a particular syndrome, it unfolded into a complex scenario involving adenocarcinomas with multiple peritoneal ramifications. This indicates the necessity for thorough and careful consideration in medical diagnoses, emphasizing the deceptive nature that can accompany initial perceptions.

Keywords: Demon Meigs Syndrome, ovarian serous adenocarcinoma.

Deciphering Gastritis: Exploring Autoimmune Roots of Anemia

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Introduction
Chronic atrophic autoimmune gastritis (CAAG) is a disease characterized by immune-mediated destruction of gastric mucosa. The aim of this case report is to highlight the association between CAAG and latent B12 deficiency, leading to pernicious anemia.

Case report
We present a case of a 52-year-old woman experiencing persistent dyspnea, fatigue, and stable angina. Upon examination, the patient exhibited generalized pallor, hypotrichosis, glossitis and hyperpigmentation on her palms and soles. Additionally, she had a long-standing history of vitiligo. Blood tests showed high LDH, pancytopenia with RBC 1,13 million/mm^3, WBC 1890/mm^3, platelets 48.000/mm^3, Hgb 4,6 g/dl, MCV 126 fl, MCH 41,2 pg and reticulocytosis. Serum vitamin B12 level was <50 pg/ml, with folate in normal range. The findings on the peripheral blood smear showed anisopoikilocytosis, erythrocytes with Howell-Jolly bodies and hypersegmented neutrophils. Thyroid ultrasound raised suspicion of autoimmune thyroiditis , confirmed by low serum FT4 and high TSH. Upper gastrointestinal endoscopy showed thinning of the stomach lining, flattened rugal folds and visible submucosal vessels. The stomach biopsies revealed atrophic gastritis, histological changes included glandular hypotrophy, fibrosis, and inflammatory cell infiltration. Anti-intrinsic factor antibodies were positive. The patient was initiated on parenteral vitamin B12, resulting in notable improvements such as normalization of the complete blood count and amelioration of digestive and neurological symptoms.

Discussions
The diagnosis of pernicious anemia relies on low serum B12 levels, positive anti-intrinsic factor antibodies and evidence of CAAG through endoscopic and histological findings. Untreated, vitamin B12 deficiency associated with CAAG increases the risk of gastric cancer and improper function of the exocrine glands of the stomach.

Conclusions
This case study reveals the link between CAAG and pernicious anemia, often accompanied by other autoimmune diseases. Early intervention is crucial, highlighting the risk of gastric cancer and iron deficiency due to hypochlorhydria. Due to the risk of malignancy, it is imperative to undergo careful monitoring.

Keywords: atrophic gastritis, pancytopenia, pernicious anemia, B12 deficiency

Hepatocellular carcinoma after direct-acting antiviral hepatitis C virus therapy: A matter of dabate

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Background
Hepatocellular carcinoma (HCC) is the most prevalent histological type of primary liver neoplasm, with chronic hepatitis C virus (HCV) infection identified as a significant risk factor for its development. Despite progress in the treatment of HCV infection, represented by oral direct-acting antivirals (DAAs), there is still a controversy about the occurrence of HCC after HCV eradication.

Presentation cases
We present the case of a 71-year-old male patient with chronic viral hepatitis C, successfully treated with DAAs three years ago, who presented to the emergency room for an enlarged abdomen, jaundice, and loss of appetite. On physical examination, the patient had jaundice with pruritus, pain in the right hypochondrium and vulgar psoriatic lesions in the abdomen and posterior chest. The laboratory analysis showed marked syndrome of hepatic cytolysis (alanine aminotransferase (ALT) = 159 U/L, aspartate aminotransferase (AST) = 733 U/L), icteric cholestasis (alkaline phosphatase (ALP) = 313 mg/dL, gamma-glutamyl transferase (GGT) = 506 U/L, total bilirubin = 8.11 mg/dL, conjugated bilirubin = 7.54 mg/dL), inflammatory syndrome (total number of leucocytes = 17.070/µl, C-reactive protein= 18.92 mg/dl) and an impaired renal function (urea = 72 mg/dl, creatinine = 1.97 mg/dl). A computed tomography scan was conducted to examine the abdominal and pelvic region. The results revealed the presence of multicentric hepatocarcinoma along with right portal vein thrombosis. Having an Eastern Cooperative Oncology Group (ECOG) performance status 3 and HCC Barcelona clinic liver cancer (BCLC) D, the patient was directed to the palliative care service.

Discussions
Although successful treatment of chronic HCV by DAAs has been confirmed by many studies and by the current clinical practice, the occurrence of HCC after SVR is a subject of debate.

Conclusion
In conclusion, the treatment plays a pivotal role but also, the follow-up of the patient is crucial for correct management.

Keywords: hepatocellular carcinoma; chronic viral hepatitis C; oral direct-acting antivirals;

Challenges in the diagnostic and treatment of scleromyositis

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Introduction
Systemic sclerosis and inflammatory myositis, both debilitating diseases with shared clinical manifestations, converge into a distinct clinical entity, known as scleromyositis. Overlap with systemic sclerosis occurs in 40% of autoimmune myositis cases with a marked tendency towards unfavourable progression, particularly among males.

Case report
A 59-year-old male presented with a 3-month history of symmetrical polyarthritis affecting small joints, proximal symmetric muscle weakness, subcutaneous edema, and skin thickening on his forearms and lower legs, accompanied by weight loss and delayed Raynaud's phenomenon without ulcerations. Elevated inflammatory markers (ESR=91mm/h, CRP=12.43 mg/dl) and muscle enzymes (CK, LDH) were observed, along with positive results for ANA, RF, anti-Scl70, and anti-Jo-1 antibodies. Capillaroscopy showed a nonspecific pattern, while electromyography revealed a myopathic pattern. HRCT scan demonstrated fibrotic changes, ground glass opacities, and micronodules in both lungs. Differential diagnoses of eosinophilic fasciitis and cancer were excluded. The patient was diagnosed with systemic sclerosis in overlap with inflammatory myositis, for which he received treatment with corticosteroids, Methotrexate, and Cyclophosphamide, with inadequate response. Considering the severity of inflammatory myositis and the presence of lung involvement, rescue therapy with Rituximab was taken into consideration.

Discussions
This case highlights a severe overlap syndrome in a male patient, combining systemic sclerosis and inflammatory myositis, with a high risk of severe pulmonary damage and mortality. The absence of established therapeutic guidelines contributes to the complexity of the medical management regarding this condition. The use of high-dose corticosteroids, crucial in myositis treatment, is limited due to the potential risk of triggering a scleroderma renal crisis. Initiation of biologic therapy, such as Rituximab, can be an alternative approach, with close monitoring of patient response.

Conclusion
Diagnosing and treating systemic sclerosis overlapping with inflammatory myositis poses clinical challenges. Further research is crucial to refine therapeutic approaches and improve outcomes for individuals with this complex condition.

Keywords: systemic sclerosis, inflammatory myositis, overlapping syndromes, scleromyositis

Infectious pericarditis with syncopal onset

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Introduction
Acute pericarditis is an inflammatory syndrome of the pericardium with or without pericardial effusion. Clinically, the patient presents with sharp chest pain and signs of cardiac tamponade, or in infectious pericarditis, an inflammatory syndrome.

Case report
We describe the case of a patient who presented in the emergency room of an 83-year-old elderly man who, for about 2 weeks, had repeated falls, with a short duration of about 1-2 minutes, with a frequency of 2-3 per week. From his medical history we note: chronic atrial fibrillation with medium rhythm, lacunar vascular accidents, Parkinson's disease. Treatment at home consisted of rivaroxaban 15 mg per day, levodopa/carbidopa 250/25 mg half a tablet three times a day, nicergoline 30 mg per day. During the clinical examination, the patient was afebrile, blood pressure 120/80 mmHg, arrhythmic heart sounds, muffled with a frequency of 80 per minute, extrapyramidal syndrome (rest tremor, plastic hypertension, bradykinesia), an ataxic syndrome, vertigo syndrome. The EKG showed atrial fibrillation with an average frequency of 83 beats/minute, hypovoltage QRS complexes and the laboratory found an inflammatory syndrome.

Discussions
The cranio-cerebral CT examination revealed cerebral atheromatosis, chronic lacunarism and cortico-cerebral atrophy. The chest CT examination revealed an important pericardial effusion with liquid densities, with a maximum thickness of 60 mm at the top of the cord, as well as a predominantly left basal pleural effusion with minimal subsegmental lung collapse subdiancent. No intra or retro peritoneal free fluid was evident. The pericardial puncture revealed hemorrhagic pericarditis (in a patient on anticoagulant treatment) and the microbiological examination of the liquid determined the presence of *Escherichia Coli*.

Conclusion
In conclusion, extremely poor signs and symptoms can hide a serious diagnosis in an elderly patient with multiple comorbidities, highlighted with the help of rigorous paraclinical explorations.

Keywords: Acute pericarditis, arrhythmic heart sounds, hemorrhagic pericarditis, inflammatory syndrome.

Thiazide-associated hyponatremia (TAH): a typical picture of SIADH

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Introduction

Hyponatremia, a life threatening condition, is a side effect of thiazide use in a minority of susceptible patients. Frequently, it occurs soon after the onset of the treatment, although there are cases when hyponatremia develops months or even years later. The clinical and biochemical profile of patients with thiazide-associated hyponatremia may be that of extracellular volume depletion and in a variety of cases, it may mimic a syndrome of inappropriate antidiuretic hormone secretion (SIADH).

Case report

Woman, 67 years old, with a history of cardiovascular diseases, is admitted to the nephrology department complaining of nausea, dizziness, lack of appetite, weight loss. She has been undergoing treatment with perindopril-indapamide (10mg/2.5 mg) for 8 months. Laboratory tests show low serum sodium (Na=107 mmol/L), hypokalemia (3,29 mmol/L), low uric acid levels (<1,5 mg/dL), low plasma osmolality (237 mOsm/kg) and increased urinary sodium (168 mmol/L). Total serum proteins, glucose and lipids levels, cortisol and TSH are within normal limits. After performing a CT scan of the head and chest, no abnormalities were found. On examination, she appears to be clinically euvolemic. The perindopril-indapamide treatment was withheld and the patient was started on hypertonic saline infusion, oral fluid restriction and increased sodium intake. During follow up, her sodium level along with the rest of the electrolytes became normal and her quality life and cognitive status were improved.

Discussions

Laboratory investigations are highly suggestive for euvolemic hyponatremia. In this category, diuretic-induced hyponatremia, hypothyroidism, syndrome of inappropriate ADH secretion (SIADH) and psychogenic polydipsia were the differential diagnosis. Based on her medical history, clinical and laboratory findings, significant improvement after thiazide withdrawal, the most likely diagnosis was TAH (SIADH-like).

Conclusion

Our case confirms that thiazides can induce hyponatremia and the clinical and biochemical profile consists with a SIADH. Physicians must be aware about the potential of thiazide diuretics to induce hyponatremia.

Keywords: thiazide, hyponatremia, SIADH

A Story About Not Giving Up: A Case Report of a 33-Year-Old Patient with Multiple Myeloma and Hereditary Spherocytosis

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Introduction
Multiple myeloma (MM) commonly affects older adults, with a median age at diagnosis of about 70 years. Hereditary spherocytosis (HS) is a congenital hemolytic disorder known for its spherical shaped red blood cells. Cases with young patients having both MM and HS are rare. Therefore, this case report highlights a patient with both MM and HS and the consequences of the case's challenging treatment management.

Case report
In February 2018, a 33-year-old man presented with persistent bone pain in his lower back and calves, myalgia, nocturne sweating, low fever, and unexplained weight loss. On examination, scleral subicter and splenomegaly were noted without palpable adenopathies. Following laboratory and paraclinical investigations, MM and HS were diagnosed. During many chemotherapy lines, the patient experienced recurring episodes of severe anemia: initially he underwent VCD therapy followed by autologous stem cell transplantation (ASCT) and Lenalidomide consolidation therapy. In 2020, disease relapse occurred, thus KRD salvage therapy was initiated. An early relapse prompted DVD salvage therapy, followed by Pomalidomide and Dexamethasone. Despite the treatment, the patient developed frontal plasmacytoma, thus PACE salvage therapy was given followed by his second ASCT and Lenalidomide consolidation therapy. The early post-transplant relapse with the development of mandibular and frontal plasmacytoma prompted DKD salvage therapy. The patient developed orbital plasmacytoma despite receiving treatment. PACE therapy was given, followed by Teclistamab therapy, which led to regression of the plasmacytomas and presented an excellent partial response.

Discussions
The management of severe anemia in patients with MM undergoing treatment and with underlying hematologic conditions such as HS is clinically challenging. Thus, patients with MM and HS require special treatment management to optimize patient outcomes.

Conclusions
Due to the challenges in therapy management, further studies are crucial to explore optimal treatment strategies in patients with MM and HS.

Keywords: multiple mieloma, hereditary spherocytosis, severe anemia, plasmacytoma

From Gastric Shadows to Blood Mysteries: Pseudo-Thrombotic Microangiopathy – A Rare Complication Due to Vitamin B12 Deficiency Post-Total Gastrectomy

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Introduction
The absence of gastric intrinsic factor, coupled with the altered anatomy post-gastrectomy, places individuals at an increased risk of severe vitamin B12 deficiency. Pseudo-thrombotic microangiopathy secondary to vitamin B12 deficiency is a rare hematological complication.

Case Report
This case report presents the unusual manifestation of severe anemia in an 81-year-old female patient with a history of gastric adenocarcinoma who had previously undergone total gastrectomy with eso-jejunal Roux-en-Y anastomosis in 2017. The patient presented with a six-month history of loss of appetite and fatigue. Physical examination revealed jaundice. Laboratory investigations unveiled pancytopenia with severe macrocytic anemia, mild leucopenia, and moderate thrombocytopenia, elevated total bilirubin (predominantly indirect), abnormal liver function tests, and decreased levels of albumin and total protein. Further hematologic analysis revealed increased iron, ferritin, folic acid, LDH, and reticulocyte levels, while vitamin B12 levels were significantly decreased. Peripheral blood smear demonstrated schistocytes and hypersegmented neutrophils. Anti-parietal cell antibodies were negative, ruling out pernicious anemia. Both direct and indirect Coombs tests were negative, excluding hemolytic anemia. A diagnosis of pseudo-thrombotic microangiopathy secondary to vitamin B12 deficiency was established. Despite the poor clinical presentation, the patient exhibited a favorable response to intramuscular vitamin B12 treatment, with significant improvement in both clinical symptoms and blood parameters.

Discussions
This case is unique due to the severe anemia's delayed onset, occurring six years post-gastric cancer surgery. The patient exhibited a rare complication of vitamin B12 deficiency leading to pseudo-thrombotic microangiopathy. Notably, the negative anti-parietal cell antibodies and Coombs tests distinguish this case from other potential causes of anemia.

Conclusions
This case highlights the importance of considering atypical causes of anemia in post-gastrectomy patients and demonstrates the need for a comprehensive diagnostic approach to uncover rare hematological complications associated with nutritional deficiencies. Early identification and appropriate management of pseudo-thrombotic microangiopathy can lead to successful outcomes.

Keywords: total gastrectomy; gastric cancer; vitamin B12 deficiency; anemia; pseudo-thrombotic microangiopathy.

Giant Retroperitoneal Liposarcoma – case report

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Background
Dedifferentiated liposarcoma (DDLS) is a mesenchymal neoplasm comprised of a mature adipocytic component and a higher-grade nonlipogenic (dedifferentiated) component. Retroperitoneal DDLS usually presents as a large mass, often encasing or invading nearby structures, posing significant challenges to its complete surgical removal, which is the mainstay of treatment. Adjuvant chemotherapy can lower the high incidence of recurrence and provide a survival benefit in some of these cases.

Case presentation
A 62-year-old male patient with a medical history of cardiovascular disease on long-term oral anticoagulation was referred to our hospital in December 2023 with complaints of diffuse abdominal pain and abdominal enlargement, increasing in the previous months. A CT scan showed a mostly cystic, well-delimited, 265/240/160-mm tumor in the retroperitoneum, encasing the pancreatic body and tail, inducing a mass effect on the abdominal organs in the upper left quadrant, and herniating through the hiatus. A multidisciplinary team meeting recommended surgical ablation of the tumoral formation en bloc with the body and tail of the pancreas, the spleen, and the left adrenal gland. Pathological and immunohistochemistry evaluation resulted in a diagnosis of dedifferentiated retroperitoneal liposarcoma pT4N0G3 L0V0Pn1 R2. Following surgery, 6 cycles of adjuvant chemotherapy (Ifosfamide plus Doxorubicin) were given through July 2023. Current imaging (October 2023) showed no evidence of disease.

Discussions
Chemotherapy regimens remain poorly efficient in dedifferentiated liposarcoma. However, benefits regarding a lower recurrence rate have been observed in the case of adjuvant chemotherapy but there is a need for more evidence supporting this. Given the surgical complexities of retroperitoneal liposarcomas which often lead to positive resection margins, chemotherapy can contribute to lowering the reccurence rate.

Conclusion
This case highlights the paramount significance of multidisciplinary care for retroperitoneal DDLS: curative-intent surgery should be performed whenever possible, while adjuvant therapy has the potential of improving long-term results in selected cases.

Keywords: Dedifferentiated retroperitoneal liposarcoma; adjuvant chemotherapy; local recurrence

Relapsing Polychondritis: a diagnostic puzzle

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Introduction
Relapsing polychondritis (RP) is a rare autoimmune systemic disease characterized by recurrent episodes of inflammation of cartilaginous and proteoglycan-rich tissues, such as the ear, nose and laryngotracheobronchial tree. It also presents with arthritis, ocular inflammation, audiovestibular involvement, heart valve incompetence. The onset is often insidious, which may render diagnosis very difficult at an early stage.

Case Presentation
A 64-year-old male with a history of hypertension and type 2 diabetes presents several episodes of syncope caused by third-degree atrioventricular block, for which a heart pacemaker was implanted. Four months later he develops sudden hearing loss, walking difficulties and fever. On clinical examination the patient presented swelling of the auricular cartilage, painful nose pyramid on palpation, ataxic gait. Blood tests revealed inflammatory syndrome, anemia and slightly elevated procalcitonin, negative blood cultures, negative ANCA antibodies. There were no abnormal findings on cranial, thoracic and abdominal CT scans. Electroneurography confirmed mixed sensitive-motor polyneuropathy. The suspicion of systemic vasculitis was raised and considering the cartilage involvement and negative ANCAs a diagnosis of RP was made. The patient was treated with high dose cortisone and iv cyclophosphamide pulses together with iv antibiotics, with favorable response.

Discussions
The diagnosis of RP is made in the presence of at least 3 clinical manifestations that prove the affection of the cartilage tissue. The disease can cause cardiovascular complications such as conduction abnormalities, which were the patients' first manifestations. Usually, the cardiovascular involvement occurs later in the disease course. This was an unusual form of presentation, and, in the absence of other symptoms, not considered to be caused by an autoimmune pathology initially.

Conclusion
RP is a rare disease, with approximately 3,5 cases per million. It can present with several symptoms and there are no specific tests, which can delay the correct diagnosis and the beginning of treatment.

Sialuria - a rare genetic metabolic disease, first case reported in Romania

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Introduction
Sialuria is a rare inborn error of metabolism. It is characterized by urinary excretion of sialic acid (>1 g/day). The impaired metabolism is caused by a deficient enzime :UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine, which is the rate-limiting enzyme in the sialic acid synthesis pathway. The main signs and symptoms caused by this pathology are delays in development, hypotonia, abnormal facial features, seizures and demyelination lessions.

Case report
We are presenting the case of a one year old boy, with no family history for inherited genetic disorders, originating from a nonconsanguineous, naturally conceived pregnancy(G2). Prenatal ultrasound markers pointed to a non-specific developmental pathology due to a 7.9mm nuchal edema spoted in week 13 of pregnancy. Furthermore in week 16 a atrioventricular canal defect was suspected, and amniocentesis with chromosomal microarray analysis was recommended. The result highlighted a normal male molecular karyotype. The pregnancy progressed without issues, though the mother had trouble feeling the fetal movements in the last trimester. After birth, the boy presented with hypotonia along with delays in achieving motor functions specific to his age. He was then directed to a neurology specialist and extensive exome testing (WES) was recommended .

The analysis identified a homozygous (bialelic) c.700+2T>C mutation in the SLC17A5 gene, classified as a variant of uncertain significance (VUS). This molecular defect has a low frequency in the general population, and algorithms for predicting pathogenicity suggest a detrimental effect on the phenotype.

Discussions
Considering that the patient is a homozygous mutant for the identified molecular defect and exhibits a phenotype suggestive of this genotypic association, further investigations are needed to determine the characteristic phenotypic spectrum.

Conclusions
It is still difficult to diagnose developmental abnormalities that are not specific during pregnancy. The obstetrics team requires a multidisciplinary aproach to elucidate prenatal complex genetic disorders.

Keywords: Sialuria, Sialic acids, Genetic testing

New compound heterozygous mutations in CFTR gene- Case report in a young adult with Cystic Fibrosis

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Introduction

Cystic fibrosis or mucoviscidosis is an autosomal recessive inheritable genetic disorder caused by heterozygous or compound heterozygous pathogenic mutations in the CFTR gene. Loss of function mutations determine the synthesis of an altered Cystic Fibrosis Transmembrane conductance Regulator glycoprotein, with impaired Cl^- and subsequent Na^+ transport and exchanges leading to viscous mucus synthesis and various phenotypical manifestations and complications mostly in the lung and pancreatic tissues.

Case report

We report the case of a 28-year-old woman with no familial history for genetic disorders who suffered from bronchiectasis of the bilateral superior lobes, partially controlled bronchial asthma with multiple infectious and allergic exacerbations, also chronic rhinosinusitis, nasal polyposis. She had been treated for right apical pachypleuritis and had post-TBC sequelae. The multiple lung infections oriented the doctors to the genetic or metabolic aspect of the patient's condition. Sweat test was performed with a 59 mmol/L intermediate value and single gene CFTR NGS testing was recommended. The molecular analysis identified 2 variants: G542X classified as a rare pathogenic mutation and 2789+5G>A, a variant of uncertain clinical significance due to low frequency and available data in literature, but for which the in-silico algorithms plead for a pathogenic, loss of function variant. Considering the patient's phenotype and molecular analysis result, she was diagnosed with G542X/2789+5G>A CFTR compound heterozygous Cystic Fibrosis.

Discussions

More than 70% of CF patients are homozygous for DF508 CFTR and have typical lung and pancreatic manifestations. The particularity of this case is the association of 2 rare variants in CFTR gene with late-onset lung related symptoms but no pancreatic manifestations. Since various genotype based targeted therapies are currently available, molecular diagnosis remains the golden standard evaluation in all patients with clinical suspicions of CF.

Conclusions

Although most cases of CF are early congenital diagnosis, compound heterozygous rare CFTR genotypes could be associated with atypical phenotypes that require particular clinical management.

Keywords: Cystic Fibrosis, CFTR, NGS, Compound Heterozygous

Multidisciplinary Approach to Rectal Adenocarcinoma with Hepatic Metastases and KRAS Mutation

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Introduction
Colorectal cancer ranks as the third most common cancer and the second leading cause of death according to the World Health Association (WHA). It is frequently associated with genetic mutations such as KRAS and BRAF. KRAS mutation can impact resistance to standard treatments and affect patient prognosis. Capecitabine, an effective chemotherapy agent used in digestive cancers, may be associated with the risk of cardiac disorders.

Case report
A 76-year-old patient was diagnosed in December 2022 with high-grade rectal adenocarcinoma, detected during a colonoscopy. Subsequent investigations revealed hepatic metastases and the presence of KRAS mutation. Oncologic treatment with Capecitabine and Oxaliplatin was initiated. After the first cycle, the patient experienced an acute myocardial infarction initially interpreted as an adverse reaction to Capecitabine.

Discussions
Colorectal cancer, often associated with rectal bleeding and anemia, poses challenges in establishing thromboprophylaxis, and KRAS mutation is significantly associated with increased mortality and thrombosis in these patients. Factors such as elevated expression of heparanase and tissue factor (TF) in malignant KRAS mutant cells may contribute to the increased risk of thrombosis associated with this mutation.

Conclusion
This case highlights the complexity of managing patients with rectal adenocarcinoma, requiring careful consideration in balancing oncologic treatment and managing the cardiovascular risk associated with KRAS mutation and the use of fluoropyrimidines.

Keywords: Adenocarcinoma, KRAS, myocardial infarction, capecitabine

Unveiling the Diagnostic Utility of Post-Mortem Imaging in VACTERL Association

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Introduction
VACTERL association encompasses a range of congenital anomalies involving vertebral defects (V), anal atresia (A), cardiac malformations (C), tracheoesophageal fistula (TE), renal anomalies (R), and limb abnormalities (L). Prenatal diagnosis is challenging due to diverse clinical manifestations and overlaps with other conditions. This abstract emphasizes the importance of post-mortem imaging in understanding fetal anomalies, improving diagnostic accuracy, and guiding management decisions in affected pregnancies.

Case report
A 30-year-old woman, gravida 2, para 1, with no significant family history, presented to the Emergency Department at 22 weeks of gestation. A transabdominal ultrasound revealed findings consistent with a posterior encephalocele in the fetus, excess amniotic fluid, and a complex cardiac malformation including an atrioventricular duct, alongside polydactyly and esophageal atresia. Following counseling regarding the potential functional prognosis, the mother and family opted for therapeutic termination of pregnancy. Subsequent autopsy findings indicated a constellation of VACTERL association anomalies, including anal atresia, left hypoplastic heart syndrome, aortic coarctation, persistent left superior vena cava, closed foramen ovale, type C esophageal atresia, horseshoe kidney, left radial agenesis, and left thumb anomaly. Additional findings included craniofacial dysmorphism, pretragian tuberculum, cleft palate, severe scoliosis, and left pulmonary hypoplasia. CT was preferred over X-ray to emphasize vertebral anomalies, revealing hemivertebrae.

Discussions
We considered syndromes with similar features, including Meckel-Gruber, Branchio-Oto-Renal (BOR), Townes-Brocks, Peters plus, and Renal hypodysplasia 3 (RHD3) syndromes. Despite shared clinical characteristics, each syndrome possesses distinctive features essential for precise diagnosis. Interdisciplinary collaboration and genetic testing are essential for confirming diagnoses and guiding patient care.

Conclusions
Post-mortem imagery enhances prenatal care by providing detailed insights into the complex nature of VACTERL association, aiding in informed decision-making and counseling for affected families.

Keywords: cardiac malformation, encephalocele, esophageal atresia

Diagnostic algorithm in CYP24A1-related Idiopathic Infantile Hypercalcemia

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Introduction
Idiopathic Infantile Hypercalcemia (IIH) is an autosomal recessive inherited cause of hypercalcemia in pediatric patients. CYP24A1 mutations have been identified as the genetic drivers, leading to an increased sensitivity to vitamin D. Rickets prophylaxis may be a trigger for severe symptomatic hypercalcemia in these patients.

Case report
A 4-month-old girl was hospitalized for recurrent vomiting, severe hypotonia, lethargy and failure to thrive, which worsened progressively in the last 2 weeks.
The clinical examination revealed: generalized pallor, decreased skin turgor, sunken anterior fontanel, tall forehead, micrognathia, high arched palate and short metacarpal bones, along with rhythmic, but weak heart sounds and a grade 2 heart murmur. The neurological exam showed weak crying and axial hypotonia.
The laboratory tests showed hypercalcemia, high vitamin D levels and low PTH and phosphate.
The renal ultrasonography detected medullary nephrocalcinosis.
Eventually, genetic testing was performed, describing a compound heterozygote state – mutations p.E143del; p.R396W – both of them loss-of-function mutations in the vitamin-D-24-hydroxylase gene, confirming the IIH diagnosis.
The rehydration and diuretic therapy normalized the calcium values and led to clinical improvement. Vitamin D supplementation was interrupted and the patient started a low calcium diet.

Discussions
The differential diagnosis of hypercalcemia can be difficult, but clinical history and examination provide diagnostic hints and guide the investigations in order to find the etiology. In this case, elucidating the underlying cause helped not only to apply the correct treatment and prevent long-term complications, but also to diagnose her sister in the presymptomatic phase of the disease.

Conclusion
Idiopathic infantile hypercalcemia should be considered everytime we encounter hypotonia and failure to thrive in infants with hypercalcemia.

Keywords: Hypercalcemia, CYP24A1, Hypotonia, Nephrocalcinosis

Navigating the Clinical Landscape of MEN 2A Syndrome: Importance of Calcitonin in Disease Management

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MEN 2A syndrome is a rare autosomal recessive genetic disease that includes medullary thyroid cancer, primary hyperparathyroidism through diffuse hyperplasia of the parathyroid glands and pheochromocytoma.

In October 2021, the patient exhibited left laterocervical swelling, which sonographically corresponded to an inhomogeneous nodule and numerous ganglion formations.

The thyroid profile was normal, but elevated calcitonin (2000 pg/ml) and ACE levels were observed. Additionally, PTH, total and ionized calcium were elevated and phosphorus was decreased. Suspecting MEN2A syndrome, a cervico-thoracic-abdominal-pelvic CT scan was recommended, revealing a left adrenal nodule. Elevated urinary metanephrines (455 ug/24 h) and plasma normetanephrines (137 ng/l) confirmed the diagnosis of pheochromocytoma.

In November 2021, laparoscopic left adrenalectomy was performed, which revealed a PASS score 1 pheochromocytoma, followed by total thyroidectomy with dissection of the lymph nodes (pT3aN1MxL1Pn1R0) and right inferior parathyroidectomy.

In February 2022, the patient underwent dissection of the left laterocervical ganglion block. Due to locoregional extension, external radiotherapy targeting upper cervical region and upper mediastinum was administered in April 2022.

Calcitonin (2674 pg/ml) and ACE (8.23) values increased continuously and in June 2022 the patient started treatment with the tyrosine kinase inhibitor – Vandetanib. Due to consistently elevated levels of calcitonin, the patient transitioned to Pralsetinib in June 2023, resulting in a significant reduction of calcitonin to 500 pg/ml.

The correlation between metastasis and calcitonin levels in MEN2A syndrome is pivotal in assessing the extent of MTC, metastatic spread, treatment response, and prognosis. Persistently high or rising calcitonin levels despite therapy indicate aggressive disease behavior and poorer prognosis, while a decline suggests a favorable response to treatment.

To conclude, managing MEN2A syndrome is a complex endeavor requiring ongoing vigilance and a dynamic approach to treatment. Given the diverse manifestations and genetic implications, there is no one-size-fits-all therapeutic solution.

Keywords: calcitonin, MEN syndrome, pheochromocytoma, thyroid

Complications of hepatic hydatid cysts: imaging approaches and management

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Introduction
Hepatic hydatid disease is a parasitic zoonosis caused by a tapeworm of the genus Echinococcus. The liver is the most commonly affected organ with an infestation rate of 60-75%, followed by the lungs, brain, heart, and bones. The aim of this particular case report is to emphasize some of the uncommon and potentially life-threatening complications, which include biliary peritonitis and secondary bacterial infection that leads to liver abscesses.

Case report
A 49-year-old man was admitted to the gastroenterology department in January 2023 with epigastric pain associated with anorexia, weight loss, as well as scleral and cutaneous jaundice. Medical history included cholestatic hepatitis and gastritis. CT and MRI showed a tumoral-like lesion in the left hepatic lobe and bilateral intrahepatic biliary dilatation. ERCP was performed revealing CHD and LHD stenosis that were treated via endoscopic stenting. Histopathological examination confirmed the biliary cyst diagnosis. In February 2023, the patient was admitted for biliary peritonitis, caused by the intrabiliary rupture of the hepatic hydatid cyst, with surgical lavage and multiple drainage being performed. In June 2023, the patients returned for further investigations, when CT scans revealed an infected hydatid cyst in the left lobe, adjacent ischemic modifications, and fluid collections, with a tendency to fistulization. The ensuing surgical interventions consisted of: aspiration and drainage of the abscesses, necrosectomy, closure of diaphragmatic fistula, as well as lavage and drainage of the remaining cavity. Pre- and post-operative administration of albendazole was initiated.

Discussions
Mebendazole was the first drug to be used for hydatid disease. Later, Albendazole was introduced due to its better absorption properties, in addition to low recurrence and complication rates of liver hydatid cysts.

Conclusions
Hydatid disease remains a continuous public health problem in endemic countries, CT being a reliable imaging method for the evaluation of uncommon but lethal complications of hepatic hydatid cysts.

Keywords: hydatid cyst, biliary peritonitis, abscesses

Laterocervical lymphadenopathy- from Basedow Graves to lymphoma: a case report

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Introduction
Diffuse large B cell lymphoma (DLBCL) is the most frequent type of non-Hodgkin lymphoma, approximately 30% to 40% of the total cases. It is mostly diagnosed in the seventh decade of life, but it can affect anyone regardless of age. An imunodeficiency is considered to be a risk factor.

Case report
This is the case of a 34-year-old female patient, known with Basedow-Graves disease, who complains about coughing and significant left laterocervical lymphadenopathy in July 2023. An ultrasound is performed and, given her medical history, she undergoes a thyroidectomy during which a biopsy of the adenopathy is performed. The biopsy report is indicating a diffuse lymphoid infiltrate, highly suggestive for a malignant lymphoma. Clinically, the cough persists accompanied by night sweating, pallor, left parasternal lymphadenopathy and jugular turgescence. The lab results indicate an isolated neutrophilia, hyperuricemia, highly elevated LDH, hyperbilirubinemia, inflammatory syndrome and elevated D-dimer levels. The immunohistochemisty confims a GCB (germinal center B-cell liketype) DLBCL, Ki67~85-90%. A TAP-CT shows a voluminous heterogeneous tumoral mass in the anterior mediastinum, extended 20.5 cm craniocaudal, 16 cm laterolateral and 7 cm anteriorposteriorly. Compression and mild right diversion of the trachea, important compression of superior cava vein, acute venous thrombosis of left brachiocephalic and internal jugular veins, and moderate left pleural effusion with tumoral infiltration, and celiac, mesenteric and periaortic lymphadenopathy were noted. Chemotheraphy protocol (R-Da-EPOCH) is initiated late September.

Discussions
Given the age and medical history of the autoimmune disease, the lympadenopathy was mistaken for a thyroid exacerbation, rather than the typical hematological suspicion which is usually high in such circumstances. The size of the tumor was unexpected and caused important deviation and compression of numerous important mediastinal structures.

Conclusions
Diagnosing DLBCL is challenging, especially in patients with complex immunologic disorders and it can be easily mistaken for other medical conditions.

Keywords: adenopathy, immunohistochemistry, compression, thrombosis

Don’t let your heart stop you from dancing: a case report on a young athlete with atrial septal defect

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Introduction
Atrial septal defects (ASD) are non-cyanogenic congenital heart abnormalities consisting of a communication between the left and the right atrium through the interatrial septum. Due to the forming of a pressure gradient, the right chambers of the heart may progressively hypertrophy leading to supraventricular arrythmias and pulmonary congestion. We present the case of a young professional dancer to illustrate the importance of early detection of congenital heart defects and the therapeutic approach in these circumstances concerning athletes.

Case Report
Our case: 14 year old boy, without any significant medical history, is investigated during a routine check-up. Physical examination: at heart auscultation was found a systolic murmur in the left parasternal intercostal 3-4 spaces with an intensity of II-III/6; normal heart sounds. The ECG disclosed one supraventricular extrasystole and further HOLTER investigation also revealed a small number (<50/24h) of supraventricular extrasystoles. A heart defect was identified by echocardiography (secundum ASD, with a left-right shunt and an incipient expansion of the right atrium). The stress test did not unveil any alteration of the exercise capacity. Periodical reevaluation was recommended, as the patient was asymptomatic.

Discussions
While physical activity lowers the cardiovascular mortality and morbidity rate in the general population, there is an increasing risk of arrythmias and sudden death caused by intense exercise in complicated cases of patients with congenital heart defects. The 2020 ESC Guidelines emphasize that asymptomatic patients without significant arrythmias, pulmonary hypertension or right ventricular dysfunction should not be restrained from practicing sports. In addition, in contrast to the overly-cautious attitude of physicians, dynamic sports have a greater positive impact on the patient’s health, compared to static activities.

Conclusions
In conclusion, the decision of limiting the physical activity of patients with ASD should be made considering and periodically revising the status of the patient.

Key words: Atrial Septal Defect, Dancer, Supraventricular Arrythmia

MOGAD Syndrome – A rare cause of autoimmunity

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Introduction
Myelin Oligodendrocyte Glycoprotein Antibody Disease (MOGAD) is an uncommon autoimmune condition where the body produces autoantibodies targeting myelin oligodendrocyte glycoprotein (MOG), a component of central and peripheral nervous system's myelin sheaths.

Case report
A 47-year-old male patient with history of chronic inflammatory demyelinating polyneuropathy (CIDP) presents to the Neurology Department for a periodic check-up. The history of the current illness includes decreased muscle tonus in lower limbs and distal paresthesia, difficulty walking, balance disorder, decreased visual acuity. The neurological exam highlights low grade paraparesis and muscular atrophy in most muscular groups found in the lower limbs, bilateral positive Babinski sign, a positive Romberg test. The electromyography identifies elements suggestive for CIDP. Visual evoked potential test is positive, showing demyelination with secondary axonal loss. A prior MRI had been done which features demyelinating lesions in the left cerebellum, pons and periventricular regions with Dawson Fingers aspect, and also at the C3 level. The lumbar puncture was not suggestive for a central nervous system infection and evidenced only an albumin-cytological dissociation which is a characteristic feature of CIDP. Blood tests had been done and were negative for any autoimmune pathology except for MOG antibodies, unraveling the etiology of all the symptoms. The patient was discharged with slight improvement of the symptoms following high doses of intravenous steroids, continuing with low doses of Medrol and immunosuppressive treatment and periodic courses of intravenous immunoglobulins.

Discussions
MOGAD is a rare autoimmune condition, and the concomitant involvement of both the central and peripheral nervous system is rare and makes this case peculiar.

Conclusions
The clinical and paraclinical findings are important for a precise diagnosis in order to receive best treatment alternatives available.

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Keywords: MOGAD, optic neuritis, transverse myelitis, CIDP

A double-edged sword: a case report on the cardiotoxic effects of cytostatic medication upon patients with preexisting cardiovascular disease

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Introduction

There is a worrying increasing number of patients diagnosed with malignancies in the past years, that also have associated comorbidities. In this context, cancer reduction treatments may induce toxicity to other organ systems unrelated to the initial neoplasm, aggravating the overall status of the patient. We present the case of an oncological patient that had an exacerbation of his cardiovascular disease (CVD) leading to general edemas after cytostatic cure was initiated.

Case report

Our case: 54 year old male, smoker, chronic alcohol consumer, was admitted to the Regional Institute of Oncology to continue the second dose of palliative treatment with Cisplatin (alkylating agent) – Paclitaxel (taxane). The patient was diagnosed with esophageal neoplasm stage IV, with pulmonary metastases. Symptoms: dyspnea, weight loss (10 kg in 2 months, BMI 17.3 kg/m^2), dysphagia for fluids and solids accompanied by edema of the lower limbs and loss of appetite. Physical examination: dullness on lung percussion and decreased vesicular breath sounds basally and bilaterally; enlarged abdomen with shifting dullness, no abdominal tenderness; clinical aspect of anasarca. Paraclinical: ECG revealed negative T waves in almost all derivations, highly suggestive for myocardial ischemia; echocardiography disclosed mitral regurgitation stage 3, moderate pulmonary hypertension and heart failure (HF) class NYHA III.

Discussions

Further clinical investigations concluded that the main cause of the exacerbated symptoms was the cardiotoxic effect of the palliative treatment and a slow, favorable evolution was observed once the medication was replaced (beta-blocker, trimetazidine, ivabradine). The 2022 ESC Cardio-oncology guidelines state that patients treated with Cisplatin, who already suffer from CVD, can develop symptomatic HF, while Paclitaxel might cause disseminated intravascular coagulation, thrombosis and HF.

Conclusions
In conclusion, the complexity of oncologic patients should make the physicians more aware when choosing the right treatment, as the balance between side effects and quality of life is crucial.

Keywords: Cardiotoxicity, Heart Failure, Edema, Cisplatin, Paclitaxel

Incidental finding of MEN1 Syndrome. A case report.

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Introduction
Multiple endocrine neoplasia type 1 (MEN1) is a rare tumor syndrome that is inherited in an autosomal dominant pattern and is characterized by the co-occurrence of primary hyperparathyroidism, duodenopancreatic neuroendocrine tumours (NETs) and/or pituitary adenomas.The prevalence of MEN1 is estimated to be approximately 1/10.000.

Case report
We present the case of a 35-year-old patient who was admitted to the GI department with a history of abdominal pain and diarrhea. Although the pacient was treated with proton pump inhibitors, the symptomatology worsened so the patient had an upper digestive endoscopy performed, which revealed multiple peptic lesions on the duodenal, gastric and esophageal walls. These findings, together with the diarrhea were suggestive of Zollinger Ellison syndrome. A SPECT-CT with Tc 99-Tektroyd was performed revealing a hyperintense nodular lesion of 18/14 mm at the caudal pancreatic level as well as other pancreatic nodules with dimensions <6 mm and a 4mm nodule in the duodenal bulb. Chromogranin A, serotonin and gastrin levels were determined, all having increased values, 3x above the upper normal limit.

Discussion
Evaluation of calcium and phosphate metabolism revealed increased PTH, increased ionic calcium, normal phosphatemia and hypercalciuria. Parathyroid scintigraphy was performed and revealed parathyroid hyperplasia, a common finding in the context of MEN 1 syndrome. The measurement of prolactin revealed increased levels and the pituitary MRI showed an inomogenous pituitary gland, with diffuse granular structure as well as a nodular lesion in the right pituitary lobe suggestive of a microprolactinoma. MEN1 Syndrome was confirmed by genetic testing (MEN1 chromosome 11 mutation: g.64571970_64571972del).

Conclusion
This case highlights the importance of imagistic evaluations in the medical practice since the patient had no symptoms suggestive of primary hyperparathyroidism or microprolactinoma. Without them, the MEN1 Syndrome would have probably been discovered much later and in a more advanced stage.

Keywords: MEN1, primary hyperparathyroidism, microprolactinoma.

Navigating Unforeseen Circumstances: Infective Endocarditis in a Previously Healthy 20-Day-Old Infant - A Case Report

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Introduction
Infective endocarditis (IE) is a noncontagious infection affecting the endocardium and heart valves, marked by thrombus formation, known as vegetation. While congenital heart disease and central venous catheters are common risk factors, late-onset GBS infection can also be a significant cause of IE.

Case Report
A 20-day-old female patient, previously clinically healthy, presented with persistent fever of 38.5°C and paraclinically, inflammatory markers were elevated (CRP= 21.3 mg/dL, procalcitonin= 97.83 ng/dL). Blood cultures were positive for Group B beta-hemolytic Streptococcus. Treatment with Ampicillin, Gentamicin, and Cefotaxime was initiated.

Transthoracic echocardiography revealed vegetations on the aortic valve (3/4.77 mm), grade 2 aortic insufficiency, grade 1 mitral insufficiency, slightly dilated right ventricle, grade 1 pulmonary and tricuspid insufficiency. Given the clinical context, NT-proBNP was measured (9922 ng/mL).

Echocardiography reevaluation showed an increase in the size of the aortic valve vegetation to 5/5.85 mm. Post-treatment, the patient became afebrile, the inflammatory syndrome decreased, but NT-proBNP worsened (52396 ng/mL), and an anemic syndrome emerged.

Diuretics were administered, and a RBC transfusion was conducted, leading to the transfer to the ICU, where the patient was hemodynamically stabilized and then transferred to the San Donato Clinic, Milano, for valve replacement surgery.

Discussions
This case highlights that one of the primary risk factors of late-onset GBS infection in neonates is maternal colonization.

Conclusions
Considering the swift advancement of the disease, prompt treatment is necessary.

Keywords: infective endocarditis, LOGBS, echocardiography

When Blood Flow Isn’t Enough- A Case Report on Peripheral Arterial Disease

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Introduction
Peripheral arterial disease (PAD) is a worldwide vascular problem where narrowed arteries limit blood flow consequently to diabetes, smoking, obesity, high blood pressure or high cholesterol, positive family history and increased age (over 50 years old). Patients with arterial narrowing due to atherosclerotic processes are unable to supply enough blood for the muscles' metabolic needs during activity, anaerobic metabolism producing metabolic acidosis with local pain, condition known as intermittent claudication (IC). Other symptoms include numb legs, coldness in the calf or foot, changes in skin color, hair loss or weak pulse. This case report aims to analyze a PAD patient's diagnosis, management and treatment.

Case Report
67-year-old non-smoker, chronic alcohol consumption male patient, known with cardiometabolic pathologies (third degree hypertension, permanent atrial fibrillation, type 2 diabetes, dyslipidemia) is admitted to the Clinical Rehabilitation Hospital for IC (pain appearing at less than 200m), coldness and paresthesia in the feet. A week prior, he was diagnosed with low limb ischemia and treated via amputation of the right second toe. His clinical examination revealed pulseless dorsalis pedis and posterior tibial arteries, and weakly pulsating popliteal arteries. The ankle-brachial index was under 0,15 (right side) and 0,30 (left side), being diagnosed with PAD stage IV Fontaine.
Further investigations showed a CT-angiography with multiple atherosclerotic processes but no intraluminal thrombi (left renal, superior mesenteric, femoral and popliteal arteries). Atherosclerotic calcifications were described in both tibial and peroneal arteries.

Discussions
PAD management focuses on maintaining an active lifestyle with required dietary changes, reducing complications development and approaching a medical or surgical treatment for preventing symptoms progression.

Conclusions
Considering the clinical and paraclinical evaluation, we have examined a patient with multiple cardiometabolic comorbidities. To improve the patient's life quality, a complete strategy involving a multidisciplinary team is required.

Keywords: ischemia, peripheral arterial disease, intermittent claudication

Case Report: Obstructive jaundice caused by locoregional recurrence of operated Gastric Signet Ring Cell Carcinoma

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Introduction
Gastric cancer is the fifth most frequently diagnosed malignant tumor in the world, acting as a significant public health problem. The Signet Ring Cell Carcinoma (SRCC) is a poorly cohesive variant composed mainly of tumor cells with abundant intracytoplasmic mucin and an eccentric nucleus, characterized by strong invasiveness and fast progression.

Case report
A 65 year-old female patient presented herself to the Gastroenterology Department for altered state and jaundice. She was diagnosed with Gastric SRCC in 2021 for which a total gastrectomy, accompanied by a Roux-en-y esophagojejunostomy and a D2 lymphadenectomy, was performed, along with radiochemotherapy. In december 2022, a colonoscopy revealed a transverse colon stenosis due to radiation colitis, treated with a jejunostomy. A subsequent exploratory laparotomy indicated the presence of peritoneal carcinomatosis, after which the chemotherapy was reinitiated. On examination, she displayed sclerotegumentary jaundice and cachexia, a distended abdomen with a post-gastrectomy xipho-umbilical scar and a feeding jejunostomy tube. Laboratory tests indicated normocytic normochromic anemia, hepatocytolysis, severe cholestasis and biliary retention. Dilated intrahepatic bile ducts and minimal ascites were found on the Ultrasound Examination. The last intervention was a percutaneous transhepatic biliary drainage by ultrasound-guided puncture of the left hepatic duct. Under treatment for hydroelectrolytic imbalances, antibiotherapy, probiotics and hepatoprotective agents the evolution was favourable, with significant decreases in biliary retention.

Discussions
This particular type of cancer has a poor prognosis due to the lack of clinical symptoms in the early stages in contrast to advanced disease stages manifested upon diagnosis, controversial findings regarding the chemosensitivity profile, locoregional recurrence and metastatic setting. Despite considerable improvements in the surgical and oncological treatments of gastric cancers, recurrences still represent an important cause of complications.

Conclusions
The patient had advanced gastric SRCC at diagnosis, and given the peritoneal carcinomatosis, the locoregional reccurence and poor functional status, she was not a candidate for further therapeutic measures. In this case, the percutaneous bile duct drainage was no more than a palliative solution for a rapidly progressive cancer.

Keywords: gastric cancer, extrahepatic cholestasis, biliary drainage

Persistent common bile duct stenosis after drained pancreatic cyst

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Introduction
Recurrent and prolonged pancreatic inflammation can lead to mass-like focal enlargement of the pancreatic head parenchyma and complications related to the compressive effect.

Case report
A 74-year-old male, with a medical history including diabetes, hypertension and cholecystectomy, was first hospitalized due to jaundice (TBil=13.4mg/dl), liver cytolysis, cholestasis and altered pancreatic enzyme levels. The patient presented a pancreatic cyst 58/63/67mm compressing the portal vein and the CBD. EUS-FNA excluded infection and malignancy of the collection, followed by drainage with a transduodenal double pig-tail catheter and an endoscopic biliary prosthesis. After drainage, the peripancreatic cyst reduced, the partially recovered patient was discharged. On reevaluation, three months later, leukocytosis and increased inflammatory markers were detected, along with liver cytolysis, cholestasis, elevated pancreatic enzymes and dyslipidemia. On ERCP, persistent intrapancreatic stenosis of CBD was observed and the plastic biliary prosthesis was successfully replaced. For E.coli positive bile and blood culture, an antibiotic course was administrated. Despite reduction of the peripancreatic collection(20mm), another cause of CBD stenosis was suspected due to inhomogeneous aspect of the head of the pancreas with inconclusive results on FNA. After 2 months, the patient was re-admitted for altered general status with nausea, vomiting and jaundice. On CT scan, dilated biliary tract, a malfunctioned biliary prosthesis and a small drained pancreatic collection were diagnosed. During ERCP, the partial migrated double pig-tail catheter was removed and the biliary prosthesis was replaced with a longer one due to 3 cm stenosis of CBD. For a cardial dilaceration, found during the procedure, 5 hemostatic clips were applied.

Discussions
The differential diagnosis and treatment of CBD stenosis can be difficult even after using all diagnostic and therapeutic tools.

Conclusions
Persistent CBD compression after a drained pancreatic cyst rises the problem of a different underling pathological mechanism in a patient with chronic pancreatitis and repeated episode of angiocolitis.

Keywords: pancreatic collection, common bile duct stenosis, mass-forming chronic pancreatitis

Differential Diagnosis Challenges in Schizophrenia

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Introduction
Schizophrenia is a heterogenous clinical syndrome characterized by cognitive and behavioral dysfunctions, along with the presence of delusions, hallucinations or disorganized speech which cause impairment of one or more major areas of functioning.

Case Report
A 43-year-old male patient with a documented history of alcohol abuse and alcohol-induced psychotic disorder was admitted to an in-patient psychiatry clinic for psychomotor agitation, verbal aggression towards the members of a Christian Pentecostal community and delusions of a mystical and persecutory nature.
Upon initial examination, the patient presented with alcoholic halitosis, psychomotor unrest, was uncooperative and showed aggression towards the medical staff.
A psychiatric exam was conducted which revealed: previous subjective episodes of depersonalization, delirious ideas of being followed and prejudiced, subjective experiences of having supernatural abilities, suspicion when being shown arguments against said abilities and high levels of irritability. Although the patient denied any suicidal tendencies, he argued that he needs to be subjected to a self-sacrifice ritual in the context of mystical delusion.
Using the DSM-5 diagnostic criteria for schizophrenia syndrome disorder, the patient's initial diagnosis was re-evaluated and the possibility of schizophrenia was taken into account.

Discussions
Alcohol induced psychotic disorder and schizophrenia both present with similar clinical characteristics, but the diagnostic criteria act as helpful tools to clinicians in the differential diagnosis process.

Conclusions
Substance abuse can trigger the onset of schizophrenia, worsen the clinical course of the disorder and the patient's adherence to treatment. Great care must be taken when examining a patient with a history of both alcohol abuse and delusions or hallucinations.

Keywords: schizophrenia, alcohol-induced psychotic disorder, delusions, diagnostic criteria.

Concurrent Lumbar Spinal Stenosis and Pulmonary Tuberculosis: A Therapeutic Challenge

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Introduction

Lumbar spinal stenosis (LSS) is a clinical syndrome of the lower extremity and back pain that dramatically reduces the quality of life, mobility and function. LSS has become one of the conditions seen most frequently in orthopedic and neurosurgical practice.
Additional conditions such as active pulmonary tuberculosis transform a regular case into a therapeutic challenge.

Case Report

Disease onset occurred insidiously 3 years ago in a 39-year-old man presenting pain in the right thigh with radiation to the lumbar spine and asthenic-adynamic syndrome.
In January 2023, a lumbar spine MRI was performed, revealing sagittal plane static disorder. At the L3-L4 level, a posterior median and left paramedian disc protrusion is described, causing severe spinal canal stenosis with an approximate diameter of the dural sac of 3 mm.
Due to the aspect described on the chest X-ray, a CT scan was performed, detecting a consolidation with atelectatic component at the level of the right upper lobe and a polycyclic nodular hyperdensity with air lucencies at the origin of the segmental bronchi. The structural appearance suggests a diagnosis of reactivated tuberculosis with bronchial dissemination. Microbiological examination confirms the presence of Koch's bacillus.
The patient consequently underwent treatment with Rifampicin, Isoniazid and Pyrazinamide. The surgical approach for LSS had to be delayed.

Discussions

The concurrent presence of pulmonary tuberculosis with LSS implies an analysis of the benefits and risks of surgical and conservative treatments. The unexpected evolution of LSS that might affect elective cases, the spread of tuberculosis to other patients and theater staff and the interactions between anti-tuberculosis medication and anesthesia drugs are some of the factors that might be considered in the clinical pathway selection.

Conclusion

Therapeutic decisions regarding a prevalent diagnosis as LSS in the context of associated pulmonary tuberculosis impose a multidisciplinary approach to decrease the hazard of tuberculosis spread and clinical advancement.

Keywords: pulmonary tuberculosis, lumbar spinal stenosis, therapeutic decision

The diagnosis and treatment of chronic lymphocytic leukemia suffering Richter transformation

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Introduction
Chronic lymphocytic leukemia (CLL) is a malignancy characterized by increased production of mature but dysfunctional B lymphocytes. In some rare cases CLL can suffer Richter transformation, a complication with a dismal prognosis in which the original clone transforms into a diffuse large B-cell lymphoma.

Case report
56-year-old male presents to the Hematology Clinic of the IOCN with multiple lymphadenopathies formed 2 years ago, which are mobile and painless. A CT scan indicates the presence of swollen lymph nodes in the chest and abdomen. Blood tests, a blood smear and histopathological examination all show a lymphoproliferation with pleomorphic cells and atypical nuclei. After these investigations, the final diagnosis of chronic lymphocytic leukemia is given and treatment with Obinutuzumab (an anti-CD20 monoclonal antibody) and Venetoclax (a BLC-2 inhibitor) is initiated. The chemotherapy lasts from June 2022 to July 2023, when the patient was considered in remission. The patient comes back on February 1st 2024 presenting with lymphadenopathies. In the months between the end of treatment and this appointment he has had multiple checkups where all tests were normal. Biopsy examination shows a malignant proliferation of large lymphocytes with immunohistochemistry markers typical for an aggressive large B cell lymphoma developed in the context of CLL (Richter transformation). Following the diagnosis, the patient is treated with Polatuzumab-Rituximab-CHP.

Discussions
The initial chemotherapy was efficient in treating the CLL and because the patient was closely monitored, the unusual Richter transformation was observed in early stages. This made possible the treatment, using the newest approved medications for large B-cell lymphoma.

Conclusions
Richter transformation, even though it rarely happens, is one of the most dangerous complications of CLL. Due to recent developments in chemotherapy, we can now treat patients who experience this with specific medications that target the immunophenotype of the malignant B cells in each individual.

Keywords: Lymphadenopathies; Chronic lymphocytic leukemia; Richter transformation; large B cell lymphoma; chemotherapy

Onset of multiple sclerosis at a young age

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Introduction
Multiple sclerosis (MS) is a disease that affects the central nervous system, characterized by autoimmune inflammation and neurodegeneration. It typically presents in individuals between the ages of 20 and 40. Nonetheless, 2-5% of patients experience their initial demyelinating episodes before reaching 18 years, these episodes are marked by severe flare-ups and a rapid decline in neurological function.

Case report
A 19-year-old woman presents with an episode of sudden blurred vision in her left eye, for which she was treated with cortisone and fully recovered in 2017. Two years later, in 2019, she experienced a second episode characterized by sensory disturbances in the left side of her face and left upper limb. Following these events, she was diagnosed with relapsing-remitting multiple sclerosis(RRMS). Over the next five years, she underwent three additional episodes, including hypoesthesia in the right side of her face, and a transition from hypoesthesia to anesthesia in the left side. MRI scans revealed white matter lesions in the fronto-temporo-parieto-occipital regions, bilaterally, with perpendicular periventricular lesions, and also in the pontine, bulbar, and cerebellar areas, all without contrast enhancement. Additionally, spinal lesions were identified at the C2 and C6-C7 levels, marked by a hyperintense band. Neurological examinations showed spasticity in the right lower limb, hypotonia in the left lower limb, hyperreflexia, muscle weakness with MSS 4/5 , bilateral positive Babinski sign.

Discussion
This case is notable for the early onset of multiple sclerosis at the age of 12, along with several episodes of recurrence. Despite experiencing five relapse episodes, the individual shows only minimal to moderate alterations upon neurological examination.

Conclusion
The case emphasizes the significance of accurate treatment selection for early-onset multiple sclerosis, aiming to minimize disease progression and improve prognosis. Proper management can substantially enhance the patient's quality of life and functional autonomy.

Keywords: multiple sclerosis, RRMS, early onset, hypoestesia

Rare association between Retinitis Pigmentosa and Neurofibromatosis Type 1.A case report

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Introduction

Neurofibromatosis Type 1(NF1) is an autosomal dominant(AD) condition caused by NF1 gene mutation on chromosome 17q11.2 encoding neurofibromin,a tumor suppressive cytoplasmic protein. Lack of neurofibromin translates to excessive cell growth and accelerated tumorigenesis. Lysch nodules (LN) are iris hamartomas,being common ocular manifestations of the disease.Possible origin derived from pigmented cells, akin to neurofibromas,is still questionable.Association between NF1 and Retinitis Pigmentosa(RP), an AD inherited disease affecting rod photoreceptor system leading to progressive retinal degeneration,is not commonly seen.

Case Report

We present the case of a 54-year old male pacient admitted in the ophthalmology department,with a past medical history of NF1 and a suspected diagnosis of RP.Pacient presented multiple cutaneous neurofibromas.Anterior pole of the eye examination revealed LN on both eyes.Visual acuity test result was 1.0.Fundoscopic examination identified thinner arteries accompanied by peripapillary atrophy,with a cup to disc ratio of 0.8,and also pigmentary deposits,bone spicules shaped, in the peripheral retina.Central retinal region was not affected by pigmentary deposits,standing for the diagnosis of RP.An optical coherence tomography(OCT) of macula showed cystoid macular edema on the right eye,as a complication of RP.

Discussion

A native and contrast substance added cranio-cerebral MRI was performed,showing a left temporal lobe tumoral growth,with a 13-14 mm diameter and central contrast absorbtion,pleading for a glioma,in NF1 initial diagnosis context of the pacient.MRI showed no tumoral evidence at orbital level and no modiffications of the optic chiasms or optic nerves. Intravitreal injections with Eylea(anti-VEGF agent) were performed for reducing macular edema.

Conclusion

Discovering RP in a pacient with a past medical history of NF1 is not commonly seen among associations of multiple AD inherited diseases.Pigmentary cells possible origin of the LN can be considered an onset of the correlation between these two AD diseases.Ophthalmological examination plays an important role for determining eventual complications of progressive loss of visual field and pigmentary cells accelerated distruction.

Keywords: Neurofibromatosis Type 1; Retinitis Pigmentosa;Lysch Nodules;Pigmentary Deposits

Challenges in the management of melanoma

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Introduction
Nodular melanoma is one of the most aggressive subtypes of melanoma, characterized by a rapid growth rate. It is typically diagnosed at an advanced local stage. It often does not fit into the classic clinical criteria of ABCD. Various types of metastases are common with this type of melanoma.

Case report
A 64-year-old patient, diagnosed with nodular malignant melanoma in the lumbar region (pT4Bn1Mx) in 2015, underwent excision and adjuvant therapy with Interferon.

In 2017, he presented with headache, projectile vomiting and abdominal pain. Computed tomography detected a hyperdense hemorrhagic lesion in the right temporal fossa suspicious for metastasis. Abdominal metastases were found in the spleen, right adrenal gland and peritoneum.The patient underwent surgery with a right temporal bone flap and complete excision of the formation. After testing positive for the BRAF V600 mutation, treatment with Dabrafenib was initiated.

In 2018, upon reevaluation with CT, a tumor formation was detected in the ascending colon. Right hemicolectomy was performed, and histopathological examination confirmed metastasis from malignant melanoma.

Radiation therapy was administered to the lesion in the right temporal lobe concurrently with immunotherapy. Thickening of the jejunal wall with suspected metastasis and multiple mesenteric lymphadenopathies led to segmental enterectomy.

He followed sequential immunotherapy and targeted therapy anti BRAF. He is currently in complete remission, without treatment. During the course of treatment, the patient presented with grade III anemia.

Discussions
Diagnosis of melanoma often occurs at an advanced stage, necessitating aggressive treatment, with a heightened risk of neurological complications and deterioration in quality of life. The anemia might also be a complication.

Conclusions
Melanoma is a disease that evolves with various metastases and should be detected as early as possible through screening to avoid dire consequences.

Keywords: Melanoma, BRAF, right hemicolectomy, cerebral metastases, immunotherapy

A breathtaking case: Subglottotracheal Adenoid Cystic Carcinoma in a 16-Year-Old Female

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Introduction
Cystic adenoid carcinoma (ACC) is a malignant tumor originated from exocrine glands and represents only 1% of head and neck cancers. Subglottotracheal localisation is rare in the pediatric population.

Case report
A 16-year-old female presented to the ENT Emergency Service with respiratory failure. Progressive and persistent shortness of breath and stridor were reported over the last week.The clinical examination indicated tachypnea and use of accessory muscles.
Direct laryngoscopy revealed a large subglottic mass, without visualization of the first tracheal ring, with important narrowing of the airway and free vocal cords, with preserved mobility. There was a normal appearance of the supraglottic and glottic area.
Emergency tracheostomy to secure the airway was performed before referring the patient for a CT scan. During the surgery, the patient developed a spontaneous left pneumothorax, treated by pleural drainage.
A CT scan was performed in order to characterize the tumor, the intraluminal stenosis and the invasion of the thyroid lobes. The patient had no metastases or loco-regional recurrences.
The histopathological examination confirmed the diagnosis of cystic adenoid carcinoma. The tumor had a solid growth pattern. This pattern is associated with a more aggressive clinical course and a higher risk of metastasis. The tumor has a pathognomonic "swiss cheese" appearance due to the presence of pseudocysts.
The treatment included six months of proton therapy and supportive care. The tracheal stoma was closed at the end of treatment.

Discussion
Radiation therapy and chemotherapy are not recommended in pediatric patients. These procedures are associated with a higher risk of long-term side effects.

Conclusion
After 2 years of follow-up in our center, there were no reccurance or side effects from the treatment.

Keywords: adenoid cystic carcinoma, subglottotracheal, pediatric patient

Protein-losing enteropathy in a child with severe anemia: a case report

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Introduction
Protein-losing enteropathy (PLE) is a rare gastrointestinal complication stemming from a diverse array of intestinal disorders that compromise mucosal integrity. PLE is characterized by excessive protein loss into the gastrointestinal tract, resulting in hypoproteinemia and generalized edema. Iron deficiency (ID) stands as the prevalent nutritional deficiency among children and manifests as hypochromic microcytic anemia.

Case Report
A 12-month-old boy presented with periorbital and lower limb edema, anorexia and diarrhea, following an acute episode of bronchiolitis with Influenza A virus. Paraclinical investigations uncovered low total serum proteins (3.4 g/dl), low serum albumin (2.7 g/dl) and systemic inflammatory syndrome (CRP 5.9 mg/dl), while nephrotic syndrome was infirmed, which supports the diagnosis of PLE. Moreover, severe hypochromic microcytic anemia was discovered (hemoglobin 6.1 g/dl, HEM 21.1 pg, ferritin 3.4). Albumin was administered parenterally, and iron therapy was continued. The investigations (pediatric immunoglobulins panel) lead towards an allergic etiology of the enteropathy, most likely food protein induced, therefore a special milk formula was administered, with a considerably favorable evolution.

Discussion
The clinical presentation of PLE exhibits considerable variability depending on the underlying cause, predominantly manifesting as generalized edema due to hypoproteinemia. Severe microcytic anemia associated with PLE in pediatric patients has rarely been reported. Many clinicians may be less aware of this association when evaluating potential causes of PLE and edema in children.

Conclusion
PLE-associated iron deficiency should be considered in patients that present with generalized edema. Treatment of PLE primarily involves maintaining nutritional status through a high protein diet supplemented with fat-soluble vitamins. Alongside dietary adjustments, addressing the underlying cause and providing supportive care to mitigate complications of edema are essential.

Keywords: Protein-losing enteropathy, Hypochromic microcytic anemia, Food protein induced enteropathy

Recurrent Miscarriages in a Couple with Unique Genetic Profiles: A Case Report

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Introduction
Infertility and recurrent miscarriage syndrome (RMS) remain challenging issues that affect 1 in 4 couples worldwide, while in half of them the cause cannot be identified. Recurrent miscarriage, characterized by two or more consecutive pregnancy losses before 20 weeks gestation, occurs in about 1-2% of couples attempting to conceive.

Case report
A 37-year-old female patient with no familial history of genetic disorders presented to the clinic after two suspended pregnancies within the first trimester. Genetic investigations revealed several factors that contributed to her reproductive challenges, particularly variants related to thrombophilia: Haplotype EPCR (low risk), FV H1299R heterozygous genotype (involved in clotting disorders), PAI-1 genotype 5G/5G (with an impact on fibrinolysis) and two MTHFR variations, associated with fetal development issues.

Moreover, her partner’s constitutional karyotype showed a pericentric inversion of chromosome 9: 46,XY inv(9). Although usually associated with a normal phenotype, this abnormality has been reported more frequently in couples with infertility and recurrent spontaneous abortions.

Discussions
Recurrent miscarriage is usually associated with chromosomal abnormalities, hormonal and anatomic abnormalities, but approximately 55% to 62% are due to blood coagulation protein or platelet defects. According to the genetic profile, the patient has a moderate-low risk for thrombotic events. In this case, prophylaxis of thrombosis and neural tube defects associated with the MTHFR variation should consist of the administration of folate in the methylated form (metafolates), since folic acid is less effective. After obtaining pregnancy, amniocentesis and prenatal genetic diagnosis are suggested if NIIPT is suggestive of genetic anomalies.

Conclusions
In summary, this report sheds light on the delicate balance between genetic variants, thrombophilia, and fertility. The patient’s unique genetic profile, including heterozygous variants in MTHFR, FV, and PAI-1, highlights the need for personalized management, while the partner’s pericentric inversion of chromosome 9 adds further complexity to the case.

Keywords: recurrent miscarriages, thrombophilia, MTHFR, chromosome 9 inversion

Severe chronic kidney disease: A journey through infectious triggers

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Introduction
Chronic kidney disease represents a major problem due to complications affecting different systems: secondary hyperparathyroidism, anaemia, atherosclerotic cardiovascular disease, acid-base and hydroelectrolytic imbalances, or weakened immune response.

Case report
A 62-year-old female was transferred to nephrology department with severe azotemia and metabolic acidosis. Her past medical history included right kidney nephrectomy, chronic kidney disease, KDIGO stage G4, left kidney lithiasis and nephrostomy, colon and cecum carcinoma with permanent colostomy. The diuresis was present and laboratory examination showed moderate hypochromic microcytic anaemia, inflammatory syndrome, elevated calcium, decreased phosphorus, and PTH=113ng/mL. On the infectious side, she had a purulent collection inside the colostomy which extended to the rectus abdominis muscles, contaminated with Enterococcus faecium and Candida, and an abdominal drainage was performed. Antibiotic therapy was initiated with Ceftriaxone. The nephrostomy was changed but became infected shortly after, leading to sepsis. Uroculture and hemoculture were positive for Klebsiella pneumoniae, and empiric treatment with Meropenem was initiated, followed by the addition of Ertapenem after an infectious disease consultation. There was noted a change in neurological exam during treatment: the patient presented dysarthria, diplopia, and a slightly confused state. Secondary encephalopathy due to Carbapenem treatment was considered. Moreover, there were recurrent episodes of urinary tract infections, leading the urology team to decide on left nephrectomy.

Discussion
After simultaneous bilateral nephrectomy, perioperative hypotension can occur, necessitating vasopressor infusion. It is also worth mentioning Carbapenem neurotoxicity, more common in patients with severe or end-stage renal disease. The literature shows Ertapenem toxicity and improvement after treatment discontinuation.

Conclusion
Nephrectomy can be considered a solution for recurrent urinary tract infections, especially in cases of obstruction, kidney lithiasis, or reflux, but a single kidney can maintain normal function. In the case of bilateral nephrectomy, the patient becomes dependent on dialysis, and the average lifespan is 10 years if kidney transplant is not possible.

Keywords: Chronic kidney disease, Nephrostomy, Nephrectomy, Recurrent urinary tract infections, Ertapenem treatment

Flood syndrome in a patient with liver cirrhosis

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Introduction
Flood syndrome is a rare complication of liver cirrhosis, caused by the spontaneous rupture of an umbilical hernia through which the ascitic fluid is externalized. It is reported a high mortality rate.

Case report
We present the case of a 54-year-old male patient diagnosed with ethanolic liver cirrhosis Child Pugh B (9 points) MELD-Na 20 points. Associated, he is known with grade I/II oesophageal varices, portal-hypertensive gastropathy, minimal hepatic encephalopathy type C, umbilical hernia, normochromic normocytic moderate plurifactorial anemia, moderate thrombocytopenia, secondary hypersplenism. From the level of the umbilical hernial orifice, ascites fluid was externalized (Flood syndrome) one week after transjugular portosystemic shunt (TIPS), indicated for recurrent hydrothorax with need for repeated thoracentesis and ascites refractory to diuretics. A surgical consultation was requested– 2 sutures were placed at this level and a tissue adhesive, Glubran, was sprayed on the wound, with minimal fluid exteriorization. It was decided to reintroduce beta-blocker and diuretic treatment– stage II (Furosemide 80 mg/day, Spironolactone 200 mg/day), with a favorable response. The patient was discharged with the recommendation to clean the wound daily with betadine dressing.

Discussions
Umbilical hernia has a prevalence of 20% among patients diagnosed with liver cirrhosis with longstanding ascites. The main complications are represented by strangulation and rupture (Flood syndrome). Management of ascites plays an important role in preventing the occurrence and recurrence of Flood syndrome. It is achieved by avoiding alcohol consumption, a low sodium diet, diuretics, therapeutic paracentesis, TIPS, liver transplant. In the case of this patient, referral for liver transplantation assessment is considered due to the dysfunction of the transjugular portosystemic shunt.

Conclusions
Flood syndrome is a rare complication of decompensated liver cirrhosis with ascites. Because of the severe complications it can cause, primary prophylaxis should be carried out first, through the management of ascites.

Keywords: Liver cirrhosis, Flood syndrome, TIPS

At first it looked like a vasovagal syncope – ended up with cardiac stimulation

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Introduction
There are plenty of causes of syncope and the differential diagnosis must be made to find the exact cause and start the best management for a patient that presents with typical symptoms, especially if there are several episodes that can lead to life threatening situations due to head trauma caused by falls.

Case report
A 17-year-old girl presented to the hospital complaining of heat and sweating, lightheaded after prolonged orthostatism and losing consciousness with spontaneous full recovery. She described having three episodes prior to this event that were almost identical with this one, the first occurring in a crowded, hot environment, and another one in the morning before breakfast.
Sinus bradycardia on 12-lead-EKG with a heart rate of 50b/min and an ectopic atrial focus causing a negative P wave was found, spontaneously converting to sinus rhythm with 60b/min.
A 24h Holter EKG noted 35 pauses without QRS complex, only with atrial conduction, each pause lasting between 2 to 4 seconds diagnosis a third degree atrioventricular block.
Permanent electrical cardiostimulation was carried out using DDD mode at a frequency of 60b/min.
Cardiac MRI with gadolinium resulted in tardive, slight contrast enhancement of subepicardial layer suggesting fibrotic changes most likely post myocarditis.

Discussions
The presentation was typically enough to be regarded as a vasovagal syncope due to the setting in which the episodes occurred. Interestingly, the patient didn`t complain of palpitations, not even during the pauses recorded by the 24h Holter, misleading at first, away from the real, conduction related cause of the syncope.
Family medical history revealed that the maternal grandmother had an acute myocardial infarction during labour, at the age of 32.
The other causes of syncope were ruled out by a complete neurological and endocrine examinations and laboratory tests for metabolic or electrolyte imbalance.

Conclusions

The particularity of the case was the conduction abnormality objectified by the 24h Holter that complicated the management of the patient, requiring a cardiac stimulator rather than conservative options that would have been useful for vasovagal syncope.

After the implant of the stimulator the patient`s evolution was great, without any episode of syncope and less dizziness.

Renal manifestations of Tuberous Sclerosis Complex: a case report

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Introduction
Tuberous sclerosis complex (TSC) is a rare genetic disease, affecting TSC1 or TSC2 gene, that manifests as a multisystem disorder defined by the development of numerous benign tumors, called angiomyolipomas (AMLs). Due to TSC, mutations in the genes cause permanent activation of the mTOR pathway, leading to tumorogenicity, cellular proliferation distributed around blood vessels and lastly is mostly manifested in hemorrage. Renal complications due to TSC can lead to chronic kidney disease (CKD) and, in severe cases, end-stage renal disease (ESRD), requiring renal replacement therapy.

Case presentation
A 35-year-old female patient known with CKD and a 10-year history of TSC, presented to Nephrology department for monthly check-up with lack of appetite, asthenia, nausea, vomiting, fatigue suggesting uremic syndrome. In 2013 an abdominal CT showed a right renal AML, giant left AML with retroperitoneal hemorrhage, indicating a mandatory left total nephrectomy. Renal AMLs can lead to complications such as local growth, mechanical pressure that is imposed on the renal parenchyma, and bleeding. In 2018, the patient was treated with Everolimus (immunosuppressant) with right kidney AML not growing further. The presence of AML on a solitary kidney, hypertension and the reduced number of nephrons precipitated the onset and progression of CKD. She developed proteinuria, possible glomerulosclerosis due to hyperfiltration on the remaining nephrons. Given all the facts, ESRD was established and a vascular access via an AV fistula was created. She was initiated in chronic hemodialysis because of severe azotemia associated with clinical symptoms.

Discussions
Population-based studies found that 48-to-80 percent of patients, between ages 15-30, with TSC have kidney complications with the presence of AMLs and progression towards renal insufficiency. In patients undergoing maintenance hemodialysis, the mortality rate and morbidity burden are markedky elevated, accompanied by a diminished quality of life.

Conclusions
Due to its infrequency, this case highlights the need to spread further awareness. Vigilant surveillance and therapeutic intervention targeting these renal manifestations are imperative to mitigate the risk of CKD in TSC patients.

Keywords: Tuberous Sclerosis Complex, Angiomyolipoma, Chronic Kidney Disease, End-stage Renal Disease, Hemodialysis

Navigating Challenges in Pyridoxal 5'-Phosphate Oxidase Deficiency: A Case Report

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Introduction

Pyridoxal 5'-phosphate oxidase deficiency, a very rare genetic disorder, causes neurological symptoms like seizures, beginning soon after birth, and developmental delays due to insufficient pyridoxal 5'-phosphate(PLP) production. Treatment involves PLP or pyridoxine supplementation for improved outcomes, highlighting the importance of early diagnosis and intervention. Anticonvulsants are ineffective, but high doses of pyridoxal 5'-phosphate are beneficial.

Case report

A, a 3-years old patient, has a genetically confirmed deficiency of pyridoxal 5-phosphate oxidase, leading to metabolic generalized epilepsy. Despite treatment, he experiences persistent seizures. Additionally, he suffers from sleep disturbances and cognitive impairments. Born via cesarean section due to a scarred uterus, he had incomplete immunizations.

Throughout his treatment, A has exhibited various symptoms, including irritability, crying, clenching of hands and feet, and rolling of eyes. He also experiences sleep disturbances, feeding difficulties, and axial and limb hypotonia. Despite adjustments to his medication regimen, including the discontinuation of Haloperidol and initiation of Levetiracetam, Frisium, and Depakine syrup, his seizures persist.

In July 2023, A presents with post-seizure state improvements in social interaction, decreased hyperkinesis, and elevated plasma pyridoxal 5-phosphate levels. However, he remains irritable with sleep disturbances. Despite increased doses of Brivaracetam and reinstatement of Nitrazepam therapy, he experiences two more seizures in October 2023.

Discussion

Despite these challenges, the neurological examination reveals improved awareness, better visual contact, and more stable independent walking. EEG shows normal sleep activity with isolated bilateral spikes. Treatment continues with a focus on managing his epilepsy, sleep disturbances, and viral infections.

Conclusion

In conclusion, pyridoxal 5'-phosphate oxidase deficiency presents a complex clinical picture, as illustrated by A's case. Early recognition and intervention are crucial in managing this rare disorder, given the ineffectiveness of conventional anticonvulsants. A's ongoing struggles

highlight the need for comprehensive management strategies targeting epilepsy, sleep disturbances, and associated comorbidities to optimize his quality of life. Moreover, his case underscores the importance of continued research and awareness efforts to improve outcomes for individuals affected by this very rare condition.

Interestingly higher incidence of males clinically affected by mutated autosomal gene

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Introduction
The ARFGEF1 gene, cytogenetically located on the long arm of the 8th chromosome, encodes a GTPase responsible for intracellular mechanisms involved in neurodevelopment. A heterozygous mutation in this gene leads to patients presenting with phenotypic manifestations such as developmental delay, impaired speech, behavioural abnormalities and, in some cases seizures.

Case report
A 6-month-old male infant is hospitalized for the evaluation of a delay in his psychomotor development. Following a neurological and psychological exam, he is diagnosed with mild mental retardation along with developmental coordination disorder and developmental expressive language disorder. Having no relevant family history and presenting with craniofacial dysmorphism, deafness and hyperopic astigmatism, he is referred to a genetics clinic where a whole exome sequencing is conducted. It is consequently discovered that the patient has a heterozygous mutation of the gene ARFGEF1 c.2917G>A. To determine the origin of the mutation, both parents' DNA is analyzed by Sanger sequencing, and it is discovered that the mother has the same mutated allele.

Discussions
Although the mutations of the ARFGEF1 gene present as clinically heterogeneous, this patient has similarities to others who are affected by the same disorder. This includes being born following an uneventful pregnancy with a physiological evolution and receiving a normal Apgar score at birth, in addition to having characteristic craniofacial features such as small, low-set ears and a high forehead. The scientific literature also mentions a currently unexplained unbalanced sex ratio, with male carriers being two to four times more likely to suffer from neurodevelopmental disorders than female carriers.

Conclusions
This case illustrates the effect a mutation in the ARFGEF1 gene can have on the neurodevelopment of an individual, as well as the potential sex-dependent incidence of this disorder, which undoubtedly requires further research.

Keywords: neurodevelopmental delay, mutation, genome sequencing

When Radiology Gets You Out of Trouble – An Initially Misdiagnosed Plasmacytoma

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Introduction
Plasmacytoma is a very uncommon blood dyscrasia, more specifically, the plasma cells turn aberrant, multiply, and produce m-protein. Solitary plasmacytoma (SP) represents an early plasma cell malignancy that is akin to monoclonal gammopathy of undetermined significance (MGUS) and multiple myeloma (MM). SP subdivides into solitary bone plasmacytoma (SBP), which occurs in the vertebrae, and extramedullary plasmacytoma (EMP). SP has three courses of progression: no marrow involvement, less than 10% clonal marrow involvement, categorised as minimal marrow involvement, and more than 10% marrow involvement, which equals the diagnosis of multiple myeloma. The treatment of choice is radiation therapy (RT), because SP are highly-radiosensitive.

Case report
A 61-year-old patient presents to a territorial hospital complaining of interscapular pain, and is given the diagnosis and treatment for intercostal neuralgia. After a few months, he complains of bilateral paresthesia. Neurosurgical exam gives the diagnosis of spastic paraparesis with the origin above T6 vertebrae. A cervicodorsal spinal cord MRI reveals a tumoral mass in the T3 vertebrae with compression, fracture, and significant stenosis of the medullary canal. Thoracic-abdomino-pelvic CT scan doesn't reveal any focal lesions or adenopathy but brings to light osteolytic lesions in T3 & T4 vertebrae.
CT-guided punch-biopsy is undergone, and histopathology exam identifies a CD138-positive, KI-67: 15% SBP. Subsequently, the patient receives RT with apparent remission of the tumour. Eight months later, the patient undergoes mandatory follow-up MRI, which raises the suspicion of tumoral tissue remnants, prompting a PET-CT request.

Discussions
Given the fact that plasmacytoma symptoms are highly non-specific (headache, dysphagia, epistaxis, general pain), but debilitating, an early diagnosis is always of utmost importance, taking into consideration the great possibility of SBP conversion into MM within 2-3 years.

Conclusion
Concisely, plasmacytoma is a rare malignancy that raises serious difficulties in its correct clinical identification, showing the paramount role of combined diagnostic and interventional radiology.

Keywords: plasmacytoma; radiation therapy; CT-guided punch-biopsy; spinal-cord MRI.

Hepatocellular carcinoma with inferior vena cava and right atrium tumor thrombosis and a hypercoagulability status

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Introduction
Hepatocellular carcinoma (HCC), the primary malignancy of hepatocytes occurs mainly in the context of chronic liver disease, being a diagnosis with bleak outcome. Approximately 1-4% of patients with HCC have inferior vena cava (IVC) and/or right atrium invasion. Malignancy is also a well-known risk factor for hypercoagulability as tumor cells can express a variety of procoagulant proteins.

Case report
A 66-year-old female known with chronic hepatitis B under treatment with Entecavir was diagnosed with hepatocellular carcinoma. At an ultrasonography multiple hypoechoic masses were found, so subsequently a contrast-enhanced ulstrasonography was performed which showed multiple hypervascular masses. The CT performed showed multiple nodular, hypodense, arterial hypercapturing images with late wash out. One of the lesions invades the IVC with extension in right atrium. The vascular thrombosis extends also inferior on all the traiect of IVC, at the level of the common iliac veins. Also, the lesions invades the right portal vein. A liver biopsy was made and histopathology was suggestive for HCC. The serum α-fetoprotein (AFP) was elevated to 9988 ng/ml (normal range < 7 ng/ml).
Shortly after diagnosis the patient presented for pain and unilateral left lower limb edema. An eco-doppler confirmed common femoral vein and popliteal vein thrombosis. It was initiated treatment with Enoxaparinum subcutaneous and subsequently it was introduced the therapy with Roteas (Edoxaban) 60 mg administrated once a day.

Discussions
HCC usually metastasizes to regional lymph nodes, lungs or bones and also has a high propensity to involve local vascular and endovascular involvement. HCC involving IVC is a challenging clinical condition which requires multidisciplinary strategy.

Conclusions
I presented a case of an advanced HCC with invasion of the IVC, portal vein, and intravascular extension to the right atrium in a patient with preexisting liver disease which was complicated with a hypercoagulability status.

Keywords: hepatocellular carcinoma, thrombosis, invasion

Pros and Cons in extensive molecular germline testing in Breast Cancer

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Introduction
Breast cancer (BC) remains the most common malignancy in women with an overall lifetime risk of 11% for this type of cancer. About 10% of cases are hereditary and therefore caused by inborn genetic defects. Molecular testing is a valuable tool for identifying high risk women, still, testing results are not always easy to interpret and use in clinical management.

Objectives
The aim of this study is to determine the prevalence of mutations classified as uncertain clinical significance (VUS).

Materials and Methods
We conducted a retrospective study to determine the prevalence of mutations classified as uncertain clinical significance (VUS) in a group of 300 women diagnosed with BC in the Department of Oncogenetics of the Oncological Institute of Cluj-Napoca, Romania that have all underwent extensive 125 gene NGS germline testing after initial oncogenetic assessment.

Results
Pathogenic mutations were identified in 37 (12,3%) BC patients. As expected BRCA1, BRCA2 , PLBB2 and CHECK2 variant were prevalent, along with a positive family history of cancer. Interestingly, 68 (22,6%) of participants carried VUS mutations in high and moderate penetrant genes.
Contrary to pathogenic mutations, recommendations for the management of VUS are not clear and focus more on clinical factors and personal and family history of cancer - breast and ovarian in particular. Genetic variants that may or may not have clinical consequence can be confusing and anxiety-provoking to patients and physicians alike.

Conclusions
Rates of VUS are relatively high and increasing, mostly in non-BRCA1 or BRCA2, and this had no impact on the therapeutic or prophylactic surgical decisions. Adherence to guidelines is extremely important to avoid unnecessary procedures .

Keywords: breast cancer, VUS, pathogenic mutations, oncogenetics

Management of a pancreatic neuroendocrine tumor using eco-endoscopy

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Introduction
Pancreatic neuroendocrine tumors (pNETs) represent a seldom encountered malignancy originating from the hormone-producing islet cells within the pancreas. These tumors comprise a subset of pancreatic neoplasms and are categorized based on the specific hormones they secrete. Examples include insulinoma, glucagonoma, and gastrinoma.

Case Report
A 47-year-old male, presented to the hospital with a myriad of symptoms including hypoglycaemia as the main symptom, fatigue, weakness, diarrhea, chest pain, and fluctuating blood pressure. His medical history was significant for metabolic syndrome, severe hepatic steatosis, mixed dyslipidemia, and insulin-dependent type 2 diabetes.
Concerned about his symptoms, clinicians ordered a series of diagnostic tests. Elevated serum amylase levels prompted a CT scan, which revealed a cystic formation within the pancreas. Further characterization via MRI confirmed suspicions of a pancreatic neuroendocrine tumor.
To obtain a definitive diagnosis, an endoscopic ultrasound-guided biopsy was performed. Results revealed a well-differentiated neuroendocrine tumor (G1) situated at the isthmus of the pancreas, more likely as insulinoma.
Given the localized nature of the tumor and the patient's comorbidities, it was opted for radiofrequency ablation (RFA) administered via endoscopic ultrasound. This minimally invasive approach offered the prospect of tumor eradication without the need for surgical intervention.
Post-procedural monitoring was conducted through contrast-enhanced endoscopic ultrasound. Encouragingly, there was no enhancement of the previously identified lesion, indicating successful tumor ablation.

Discussion and Conclusions
The case was complex due to the rarity of pancreatic neuroendocrine tumors and the necessity to rule out other pathologies for accurate diagnosis. Through a combination of imaging modalities and biopsy, a definitive diagnosis was achieved. In this case, endoscopic ultrasound served both as a diagnostic and therapeutic method, being a non-invasive, efficient, and rapid technique for tumor removal.

Keywords: pancreatic neuroendocrine tumors(pNETs), insulinoma, radifrequency ablation(RFA)

Asymptomatic metastatic lung adenocarcinoma mimicking a periosteal femoral sarcoma: a rarity within a rarity

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Introduction
Adenocarcinoma represents the most common histopathological subtype of lung cancer depicting quite a revelatory clinical presentation overlooked by hemoptysis, dyspnea and pleural effusion, especially in advanced stages. Owing to high rate of dissemination, metastasis in adrenal glands, liver and bone spine are the most frequently associated distribution patterns.

Case report
A 63-years-old male patient presented with diffuse severe swelling in the right thigh accusing deep tenderness and great difficulty in movement. The initial imaging scans indicated a malignant-appearing-mass within the femur measuring over 10cm along with an inguinal adenopathy, yet no secondary lung or liver determinations were revealed. Taking the underlying clinical and imagistic features into consideration, the primary suspicion implied a periosteal osteosarcoma, a rare bone malignancy usually forming on the surface of the lower limbs.
For a better diagnostic accuracy, a biopsy was performed, the IHC analysis unexpectedly pointing a profile compatible with a pulmonary origin as well as stating a peculiar site for a massive metastasis. Therefore, a PET-CT scan was conducted and abnormal activity was indentified among the lungs suggesting multiple mediastinal and paratracheal adenopaties. All of these examinations lead to the diagnosis of a stage 4 metastatic lung adenocarcinoma, particularly lacking any specific respiratory symptoms or noticeable pulmonary tumoral nodules.

Discussions
The uniqueness of this case is reflected in its unusual clinical description as a large femoral metastasis of a poorly symptomatic lung adenocarcinoma has been rarely reported in medical literature. Making the right diagnosis impacted notably the outcome of the disease whereas the orthopaedic surgery was declined and the proper radiation and chemotherapy treatments were added resulting in an impressive tumoral reduction.

Conclusions
Thanks to increasingly more precise imagistic and laboratory methods even strangely-located malignancies prove to be easily and correctly exposed prompting specialists to be more aware about the intricacies of well-known pathologies.

Keywords: lung adenocarcinoma, femoral osteosarcoma, metastasis

Unexpected Twist: Autoimmune Hepatitis Unmasked by Pediatric Pneumonia

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Introduction
Pediatric autoimmune hepatitis (AIH) presents as an acute or chronic progressive disease with potential long-term consequences if left unaddressed. AIH is characterized by a broad spectrum of manifestations in children necessitating vigilant management. We present a distinctive pediatric case initially presenting with pneumonia, where the diagnosis of AIH was made incidentally.

Case Report
A 10-years-old child presents to the pediatric emergency room with acute onset of fever, cough, dyspnea, tachypnea, tachycardia, hypotension, jaundice and distended abdomen. An immediate chest X-ray showed right apical pulmonary condensation and pulmonary effusion which, correlated with the inflammatory syndrome (CPR=4mg/dl), resulted in the diagnosis of pulmonary onset sepsis (procalcitonin=16ng/ml). In addition, the paraclinical findings revealed acute hepatic insufficiency with hepatosplenomegaly (ALT=953U/l, AST=849U/l, INR=2.6, APTT=52sec, TQ=33sec, Total Bilirubin=8mg/dl). The medical team transferred the patient to ICU, where the condition improved under antibiotics, before being managed in the pediatric department.

Discussions
To address the rapid disease progression in children, a comprehensive range of etiologies was explored to elucidate the underlying cause. Infectious hepatitis, alfa1-antitrypsin deficiency, Wilson disease and thyroid or surgical causes were excluded. In addition, myocarditis and LES were included in the differential diagnosis, but ruled out by further echocardiography and paraclinical panels.

However, IgG elevated titer, positive ANA antibodies=1/320 and SMA antibodies=1/40, echographic modified aspect of the liver and all the paraclinical examinations led the medical team to the diagnosis of type 1 autoimmune hepatitis, upon ESPGHAN criteria, when the immunosuppressor treatment was initiated with favorable outcomes.

Conclusions
This case highlights the importance of maintaining a broad differential diagnosis, particularly in pediatric patients presenting with seemingly unrelated conditions. The incidental discovery of autoimmune hepatitis during the evaluation of pneumonia underscores the need for heightened clinical suspicion and comprehensive evaluation. Timely recognition and management of AIH can significantly impact patient outcomes and prevent long-term complications.
Keywords: autoimmune hepatitis**,** pediatric patient, sepsis

Unveiling the silent challenge: pulmonary atresia with intact ventricular septum (PAIVS) in an unsupervised pregnancy

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Introduction
A distinct and intricate challenge in the vital domains of neonatal and pediatric heart care arises from the occurrence of tricuspid regurgitation and pulmonary atresia with intact ventricular septum (PA-IVS) in a newborn. This rare congenital heart defect, marked by the total occlusion of the pulmonary valve alongside a preserved ventricular septum and compromised tricuspid valve performance, demands prompt and sophisticated medical responses.

Case Description
Following a C-section delivery at 38 weeks due to marginal placenta praevia, a 2-day-old female newborn from an unsupervised pregnancy was diagnosed with pulmonary atresia. Within hours of delivery, the 3.1 kg baby showed signs of respiratory distress and cyanosis, which prompted the neonatal care team to assess her right away. A significant congenital heart defect was suggested by the initial physical examination, which revealed diminished pulmonary blood flow and a single, loud second heart sound. The diagnosis was confirmed by echocardiography, which also revealed tricuspid regurgitation, patent foramen ovale (PFO), and an intact ventricular septum, which distinguished PA-IVS from other forms of pulmonary atresia. In order to preserve ductal patency and guarantee a certain degree of pulmonary blood flow, prostaglandin E1 was used to stabilize the baby due to the critical nature of the diagnosis. To quickly decide on a course of action, a multidisciplinary team comprising neonatologists, cardiac surgeons, and pediatric cardiologists was quickly assembled.

Discussion
In the unstable setting of high-risk situations such as placenta praevia in an unsupervised pregnancy, this case report aims to shed light on the challenges associated with diagnosing and treating this specific subset of cardiac abnormalities.

Conclusion
This case highlights how important it is to conduct a thorough clinical examination on infants in order to detect severe congenital cardiac defects such as PA-IVS. Prompt intervention and early detection are essential for managing this condition.

Keywords: Pulmonary atresia, placenta praevia, CHD

Unraveling Complexity: A Pediatric Infection Case Report

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Introduction
Infections in pediatric patients can present with a myriad of symptoms and complications, often requiring a comprehensive approach to diagnosis and management. We present a case of a 10-year-old female patient with multiple concurrent infections, including sepsis, osteomyelitis, acute erythematous-pustular angina, and an infectious rash.

Case report
A 10-year-old female presented with fever, vomiting, diarrhea, odynophagia, and a maculopapular rash,initially treated symptomatically and with macrolides, did not recover under mentioned treatment. She developed a significant inflammatory syndrome, prompting referral. Imaging revealed pulmonary interstitial markings and hepatic steatosis, with suspicion of a congenital single left kidney. Three weeks prior, she accused a sprain and the existence of a right foot skin lesion, both untreated. Physical examination showed signs such as: raspberry tongue, upper and lower limbs edema, acute dehydration, alongside laboratory findings of elevated inflammatory markers. Serological tests excluded measles and the throat swab came back negative for streptococcus B-hemolytic and other bacterial cultures. Treatment with ceftriaxone, fluid resuscitation, and analgesics proved insufficient, leading to further investigations confirming osteomyelitis. Broad-spectrum antibiotics and corticosteroids were initiated, with subsequent improvement. Additionally, she complained of dysuria and inguinal pruritus, treated as possible candidiasis. Osteomyelitis responded favorably, prompting de-escalation of antibiotic therapy after 7 days.

Discussions
Sepsis, osteomyelitis, acute erythematous-pustular angina, and infectious rash pose diagnostic challenges and require a multidisciplinary approach. Timely recognition and appropriate management, including broad-spectrum antibiotics and supportive care, are crucial for optimal outcomes.

Conclusions
This case emphasizes the complexity of pediatric infections and the importance of comprehensive evaluation and multidisciplinary management.

Keywords: Pediatrics, Sepsis, Osteomyelitis, Multidisciplinary approach

A case of ventricular arrhythmia and Marfan syndrome

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Introduction
Marfan Syndrome (MFS) is a rare disorder with autosomal dominant inheritance in which the fibrillin gene is mutated, resulting in abnormalities of the eyes, bones, heart, blood vessels, lungs, and central nervous system. Mitral valve prolaps (MVP) is a valvular abnormality which is often associated with MFS and that causes mitral regurgitaion. Nonsustained ventricular tachycardia (NSVT) means three or more consecutive ventricular beats at a rate of greater than 100 beats/min with a duration of less than 30 seconds

Case report
We present the case of a 16-year-old female patient who had an episode of syncope, which led her to a paediatric cardiology consult. The clinical exam showed: weight deficit, height development above the average; craniofacial dysmorphism, arachnodactyly, pectus excavatum, coxa valga, dorso-lumbar scoliosis, genu valgum, holosystolic murmur grade IV with axillary irradiation. The paraclinical examination showed: mitral valve prolapse, grade III mitral insufficiency, dilated left atrium and ventricle; nonsustained ventricular tachycardia, ventricular extrasystoles, high NT-pro-BNP. Family history showed 2 cases of sudden death (father 48 years old, sister 21 years old) and past medical history included congenital cataract and lens subluxation. The patient has a Marfan Systemic score of 6. Based on all these findings, the diagnosis of MFS was made. The patient's heart condition qualifies as NYHA III heart failure. During the hospitalisation the patient was treated with amiodarone, which reduced the number of ventricular extrasystoles. A loop recorder was also implanted in order to evaluate the efficacy of the antiarrhythmic treatment.

Discussions
NSVT is recognised as a potential marker for developing sustained ventricular arrhythmias and sudden death. It is believed that the MVP can cause arrhymia through the mechanical stress induced, which leads to fibrotic changes in myocardium.

Conclusions
MFS comes with a broad spectrum of abnormalities affecting many organs and the ones of the cardiovascular's system are some of the most common and most dangerous.

Keywords: Marfan Syndrome, Mitral valve prolaps, Ventricular Arrhithmia

A Pediatric Case of Dyspnea: Navigating Hepatopulmonary Syndrome in Congenital Hepatic Fibrosis

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Introduction
Congenital hepatic fibrosis manifests as a chronic, progressive condition with enduring complications, including the development of hepatopulmonary syndrome (HPS). HPS is a pulmonary vascular disorder characterized by intrapulmonary vascular dilation leading to impaired oxygenation. This case illustrates an intriguing pathophysiological mechanism, associating dyspnea as the initiating factor.

Case report
A 10-year-old patient presents to the pediatric emergency department with recurrent episodes of dyspnea. The physical examination reveals marked peripheral cyanosis, with an SpO2 of 80%, clubbing of the fingers, scleroicterus, and significant splenomegaly. The patient has a known history of congenital hepatic fibrosis since early childhood, diagnosed through liver biopsy. Cardiology consultation indicated persistent patent ductus arteriosus and pulmonary hypertension. The presence of intrapulmonary shunts, secondary to HPS, was suspected. Microbubble ultrasound was performed, revealing bubbles in the left heart cavity between the third and the sixth beat. The calculated P(A-a)O2 gradient was elevated. A computed tomography scan confirmed an arteriovenous communication in the left lower pulmonary lobe.

Discussions
The understanding of the physiopathological mechanism is a key factor in developing a treatment. Hepatic cirrhosis and liver dysfunction lead to bacterial translocation and endothelial cell proliferation, resulting in an imbalance of local vasodilatory factors (nitric oxide) and pulmonary angiogenesis. Rapid screening and patient assessment following rigorous algorithms can assist the physician in prioritizing the patient for liver transplantation. Patients with end-stage liver disease, coupled with hepatopulmonary syndrome, have a median survival of 24 months compared to those without HPS, who have a median survival of 84 months. Ongoing research is actively investigating its pathogenesis, with a particular focus on the role of angiogenesis, aiming to identify new therapeutic targets.

Conclusions
As there is no definitive medical cure, and resolution is achievable only through liver transplantation, certain patients may be selected as a priority in the transplant list. Hepatopulmonary syndrome is characterized by gas exchange abnormalities resulting from intrapulmonary vasodilations and arteriovenous shunting.

Keywords: angiogenesis, liver transplant, pulmonary shunt

Section 3

Poster Presentations

Clinical Surgical

3.1.Winners of the competition

First Place

Single Hepatic Metastasis in Patient with Duodenal GIST: a case report

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Introduction
Duodenal gastrointestinal stromal tumors (GIST) are a rare type of malignant tumors, usually caused by mutations in KIT or PDGFR genes, resulting in abnormal tyrosine kinase receptors. The origin of GIST is considered to be the interstitial cell of Cajal, a specialised cell important for the gastrointestinal motility.

Case description
A 54-year-old woman presented with recurring intense pain in the right hypochondrium, that started 6 months ago. No other associated symptoms were described. The patient was diagnosed 4 years ago with duodenal GIST, for which surgical resection was performed and the follow-up exams were promising. On examination, abdominal distension and dullness on percussion in the right abdominal flank were noticed and on palpation, an enlarged liver was felt. A CT scan revealed a tumor of 20/18 cm in the right hepatic lobe and the histopathological exam confirmed the initial diagnosis of hepatic metastasis of the duodenal GIST. Right lobe hepatectomy, as well as a cholecystectomy were performed. She was discharged 11 days after the surgery and started adjuvant therapy with tyrosine kinase inhibitors.

Discussion
Hepatic metastases in patient with duodenal GIST are common, and are usually a sign of poor evolution. The management of the patient should include both hepatic resection and adjuvant therapy consisting in tyrosine kinase inhibitors. Combined therapy, with Imatinib and Sunitinib showed better results than monotherapy. After complete surgical resection, followed by the combined adjuvant therapy, the prognosis of the patient improves significantly.

Conclusions
Given the high risk of hepatic and extrahepatic metastases of duodenal GIST, thorough investigations should be performed periodically, as to eliminate the possibility of recurrence.

Keywords: duodenal GIST, hepatic metastasis, right lobe hepatectomy, cholecystectomy, tyrosine kinase inhibitors

Second Place

Tackling Knee Deformities: Unveiling the Importance of the Tibial Stem in Primary Arthroplasty

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Introduction
During knee arthroplasty, addressing severe deformities and bone defects remains a challenging issue. Treatment options include the use of the tibial stem and metal augments for both bone defect coverage and joint stability in the frontal and rotational planes. The employments of metal blocks often require distal augmentation with a short cemented or long uncemented tibial stem. Incorporating the tibial stem reduces the failure rate, particularly in cases of significant varus/valgus deformities with accompanying defects.

Case report
We describe the case of a 54-year-old patient with a history of multiple surgeries on the contralateral ankle, culminating in ankle arthrodesis and a secondary limb length discrepancy of 7 cm. The patient presented with left knee pain, grade III varus deformity, and relative functional impairment. Radiological examination revealed a varus deformity of 27^0 (HKA), internal femoral condyle necrosis causing femoral varus (LDFA= 98 – lateral distal femoral angle), and significant tibial varus (MPTA= 72 – medial proximal tibial angle) due to a 15mm internal tibial defect.

Discussions
The patient underwent total knee arthroplasty on the left side, utilizing a cemented prosthesis with posterior stabilization, a 100mm uncemented tibial stem, and a 15mm tibial augment. The patient's postoperative course was favorable, with the primary postoperative recommendation being the continuous use of an orthopedic boot with a 7 cm lift on the contralateral limb during walking. Therefore, employing the extension stem for the tibial component in knee arthroplasty reduces the aseptic decementation rate of the implant by decreasing mechanical loading on the surrounding bone and osseous tensions, as well as minimizing micromotion between the implant and adjacent bone, thus demonstrating potential longevity enhancement.

Conclusions
The use of the tibial stem in primary knee arthroplasty for deformities represents a crucial strategy in cases of varus and significant anatomical alterations, ensuring efficient surgical intervention and achieving durable outcomes in their treatment.

Keywords: Knee Arthroplasty, Tibial Stem, Tibial Augment, Varus

Third Place

Pathology examination: a gold standard in diagnosing gastric leiomyosarcomas

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Introduction
Leiomyosarcomas (LMS) are soft tissue sarcomas that arise from smooth muscle cells The gastric LMS is a very rare, aggressive tumor that originates in the gastric wall. Before the KIT (CD117) immunochemistry (IHC) many gastrointestinal stromal tumors (GISTs) were frequently misdiagnosed as LMS. Since the 2000's only 13 cases of gastric LMS have been reported in literature.

Case presentation
A 76-year old patient, recently diagnosed with a gastric neoplasm without a conclusive biopsy report presents to the emergency room (ER) accusing of abdominal pain, melena and persistent vomiting. She had undergone prior investigations, including an abdominal CT that showed an ulcerated mass of the antral region wall. Upon ER arrival, a gastroscopy was performed, confirming the ulcerative source of the bleeds, while also retrieving biopsies from the non-hemorraghic areas. The suspicion of GIST was disproved by the absence of immunostaining for DOG1. Upon consultation, the team opted for a partial gastrectomy with a gastrointestinal-jejunostomy. During the surgical intervention, the diagnosis of spindle cell metastasis was made through a frozen section procedure. The final diagnosis was made on the gastric surgical resection and confirmed by specific IHC reactions for LMS. Postoperative complications did not arise so the patient was discharged a few days later, awaiting follow-up.

Discussion
The particularity of the case consists in its occurrence in the muscular layer of the stomach, as LMS rarely occur in organs other than the uterus.

Conclusion
Since it is so rare, limited information is available in regard of approach. Therefore, the diagnosis mainly relies on the histopathological identification of the spindle cell neoplasm, as well as a differential diagnosis consisting of IHC staining.

Keywords: leiomyosarcoma, GIST, gastrectomy, morphology, immunohistochemistry

First Honorable Mention

Obstructive jaundice due to hepatic hilum tumor after cephalic duodenopancreatectomy – Case presentation

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Introduction
Palliative measures in patients with unresectable hepatic hilum tumors focus on reducing the grade of biliary obstruction, alleviating patient symptoms (jaundice, pruritus) and reducing the risk of other complications (cholangitis). The approaches can be percutaneous, endoscopical or surgical.

Case report
A 64-year-old woman presents at the clinic with generalized pruritus, jaundice, choluria and acholic stools. Past medical history includes a cephalic duodenopancreatectomy for pancreatic adenocarcinoma one year ago. On clinical examination, altered general status and jaundice were noted. Laboratory examinations showed elevated total bilirubin (a value of 25 mg/dL), ALP, ALT, AST and GGT. The CT scan showed a hepatic hilum tumor, without other secondary determinations, the absence of local recurrence of the pancreatic adenocarcinoma, and biliary duct dilatation. It was decided that the best course of action was surgical. A bisubcostal incision was preformed. On inspection, the hepatic hilum tumor that was invading the IVC, portal vein, right hepatic artery and the old anastomosis was noted. The old choledoco-jejunostomy was un-done and resected, with the creation of a new end-to-side jejuno-jejunal anastomosis and an end-to-end coledoco-jejunal anastomosis, with the protection of a Kehr tube. The patient had a moderately favorable evolution, with a total bilirubin of 18 mg/dL and a decrease in the liver panel enzymes on day 7 after the intervention.

Discussion
Since the tumor was invading an already existing anastomosis, other options for biliary drainage such as stenting or percutaneous drainage were not taken into account. Possible complications include fistula, anastomotic stricture or cholangitis. Given the fact that the tumor is unresectable, the prognosis is reserved.

Conclusions
Palliative measures for biliary drainage in hepatic hilum tumors are important for assuring patient comfort and preventing complications.

Keywords: Choledoco-jejunostomy, hepatic hilum tumor, obstructive jaundice

Second Honorable Mention

An unusual place for an oligodendrogioma: case report and literature review

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Introduction
Oligodendrogliomas (ODGs) are classified as grade II low grade gliomas, according to 2022 WHO guides. Those lessions are slow-growing benign tumors, that develop out of oligodendrocytes, glial cells that, akin to Schwann cells in the PNS, produce myelin for the CNS.
The ODG found in our patient had an unusual localization: the floor of the 4th ventricle and the cerebellar vermis.

Case report
The patient, a 31 y.o. male, presented to our clinic for headaches and motor disfunctions. The onset of the symptoms was insidious, beginning one year and a half prior to admission.
The contrast enhanced cerebral MRI showed an expansive lession, in the floor of the 4th ventricle and vermis, with a right paramedian cerebellar expansion. The differential diagnosis was made with a cavernoma. On examination,the patient had a GCS of 15, presented no ICP (intracranial pressure), but presented signs of a cerebellar syndrome.
GTR (gross total resection) was chosed and achieved via a median suboccipital craniotomy with a median posterior fossa approach. Extemporaneous histopathological exam confirmed the ODG diagnosis.The first day post-op CT showed no tumor remnants and no signs of ischemic lessions. Patient was discharged after 4 days and advised to return for a 3 month follow-up MRI.

Discussions
The literature indicates that using GTR for the histopathological exam leads to a better outcome than using biopsy: a greater tumor sample can be taken, improving the overall survival rate.
ODGs account for about 5% of intracranial tumors and the association with the IDH mutation and 1p19q codeletion poses the risk of rapid tumor growth and the transformtion into a high grade glioma. The average 5 year survival rate is >90% , for the 20-40 y.o age group.

Conclusions
To conclude, ODGs are rare slow-growing benign tumors. The ideal treatment remains the surgical approach, the ultimate goal being GTR.

Keywords: oligodendroglioma, gross total resection, 4th ventricle,

3.2. Clinical Surgical

Lumbar hydatid cyst in a pregnant woman - a challenging case combining neurosurgery, obstetrics and infectious diseases

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Introduction
A lumbar hydatid cyst is a rare manifestation of an infection *with Echinococcus granulosus*, a cestode that inhabits the small intestine of canids. The infection can be transmitted to humans through the orofecal route or close contact with the infected host. In the human manifestation of the disease, hydatid cysts typically occur in the liver or the lungs.

Case report
This case report highlights the surgical management of a recurring lumbar hydatid cyst in a 29-year-old female patient residing in a rural area. This patient was admitted to the Neurosurgical department on three occasions (January 2019, February 2020 and May 2022) with similar symptomatology, experiencing intermittent sciatica and paraplegia in her lower limbs due to hydatid cysts on the conus medullaris. Following each intervention, she received treatment with Albendazole. During her hospitalization in 2020, when she was also 22-weeks pregnant, she developed a severe motor deficiency in both lower extremities and urinary retention. Despite the pregnancy-related risks, she underwent successful emergency surgery.

Discussion
The recurrence of these hydatid cysts, especially during the pregnancy, proved to be a challenge for both the patient and the medical team involved. The infectious diseases department advised towards surgery, while the obstetrics department acknowledged the risks that the intervention would impose on the fetus' evolution. However, worsening symptoms necessitated immediate surgery. Thankfully, the neurosurgical team was able to save the mother's life without affecting the pregnancy. This unfortunate situation could have probably been avoided if the patient had adhered to her treatment regimen.

Conclusion
This case shows how a patient's background can impact the success of the medical act. A lack of medical education and poor hygiene standards can alter the result of an otherwise straightforward treatment, despite the best efforts of the medical team.

Keywords: Hydatid cyst, sciatica, pregnancy

From Diagnosis to Reconstruction: A Multidisciplinary Approach of Tibial Adamantinoma

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Introduction
Adamantinoma, a rare low-grade malignant bone tumor, makes up around 0.5% of all primary bone tumors, with the tibia being the predominant location. Characterized by its locally aggressive nature, adamantinoma is known to induce lytic lesions that may lead to fractures and pain, with documented instances of local recurrence following surgical resection.

Case report
We present the case of a 40-year-old woman patient who was referred with adamantinoma of the left tibia. She presented with localized pain, crepitations, pathological mobility at the level of middle 1/3 of the left tibia, and total functional deficit after she slipped on ice. Radiographies confirmed a nondisplaced diaphyseal tibial fracture, on a pathological background with osteolysis, cortex destruction, and soft tissue extension. MRI confirmed multiple extensive bone tumors extending from approximately 7 cm below the proximal knee joint to about 8 cm proximal to the distal ankle joint and highlighted the extension into the adjacent soft tissues sparing the peroneal nerve and fibula. Bone biopsy confirmed the radiologic suspicion of Adamantinoma, and PET-CT excluded the presence of distant metastasis. After a multidisciplinary team planning, we performed en bloc-wide local tumor resection followed by bone reconstruction with a customized segmental prosthesis and soft tissue restoration using a free muscular flap from the latissimus dorsi muscle. The radiologic evolution of the patient was favorable, with the patient able to fully weight-bear without crutches at 6 months and good function without signs of recurrence at 18 months.

Discussion
The preferred treatment is wide local resection, including nearby lymph nodes and limb reconstruction. Techniques like distraction osteogenesis, allo/autografts and custom prostheses help fill bone gaps. Amputation doesn't improve survival but is an option for recurring cases. Radiation and chemotherapy have limited effectiveness.

Conclusion
Despite no specific guidelines, this approach led to good functional recovery and no recurrence 18 months post-surgery, highlighting the potential success of surgical intervention in such cases.

Keywords: adamantinoma, custom implants, multidisciplinary approach

Neurosurgical treatment of complications occuring in posterior fossa stroke

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Introduction
Ischemic stroke is a neurological emergency that occurs when the presence of a blood clot in a cerebral artery leads to a reduction or interruption of blood supply to a part of the brain. This deprives the brain tissue of oxygen and nutrients, which results in the death of brain cells. Hemorrhagic transformation is a major complication of ischemic strokes that can be promoted by thrombolytic treatment in the acute phase, anticoagulant treatment (necessary for patients with risk factors in the secondary prevention of ischemic stroke or in the prevention of venous thromboembolism in immobilized patients), but sometimes it can occur spontaneously.

Case presentation
We present the case of a 79-year-old patient who presented to the Emergency Department with a motor deficit in the right limbs that had developed over the past few days, accompanied by coordination, gait, and balance disturbances. Upon admission, she was diagnosed with RIGHT ATAXIC HEMIPARESIS, CHRONIC ATRIAL FIBRILLATION, and HYPERTENSION. A cranial CT scan revealed an ischemic stroke in the left cerebellar hemisphere. The patient received treatment for cerebral edema, anticoagulants, lipid-lowering medication, neurotrophic antihypertensive medication, and vestibular therapy, with favorable progress, allowing for supported walking within the first 7 days. However, her consciousness deteriorated, and the motor deficit worsened, ultimately rendering her paralyzed with a motor score of 0/5 on the Functional Motor Scale (FMS) and a Glasgow Coma Scale (GCS) score of 6 points.

Repeat paraclinical investigations did not show significant changes, but a cranial CT scan revealed an ischemic stroke in the left cerebellum with hemorrhagic transformation. The presence of this complication required transfer to the neurosurgery department, where atlantooccipital decompression and hematoma evacuation were performed. Subsequently, the patient's condition improved with neurological recovery and without major complications.

Conclusion
The treatment of strokes is a complex issue, and patients with multiple comorbidities (in this case, an acute ischemic stroke in the context of permanent atrial fibrillation) may experience major complications. Proper monitoring and surveillance provide the opportunity for a rapid diagnosis and treatment with a favorable outcome.

Keywords: Ischemic stroke, atrial fibrillation, hemorrhage

Lymph node management in metastatic papillary thyroid carcinoma in a 29-year-old with Hashimoto's thyroiditis - a case report

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Introduction
Follicular and papillary carcinomas comprise 95% of thyroid cancers. Neck lymph node metastases are frequent in differentiated thyroid cancer (DTC). However, removing only metastatic lymph nodes does not significantly affect DTC survival and recurrence. Therefore, a systematic cervical dissection is the preferred approach.

Case report
This case report provides an overview of the treatment of a 29-year-old woman with Hashimoto's thyroiditis. She visited the endocrinology department after noticing an enlargement in her neck region. The ultrasound showed no nodules and an enlarged thyroid with heterogeneous structure. Two calcified spots in the left lobe that indicated a high probability of malignancy were present. Bilateral neck examination revealed numerous suspicious lymph nodes, prompting a native cervico-thoracic CT scan, confirming a significantly enlarged thyroid with multiple calcifications, and verifying the presence of cervical lymphadenopathy, which also exhibits calcifications. A total thyroidectomy is performed, with the specimen sent for immediate histopathological examination. The result confirms papillary carcinoma, and the procedure continues with a modified cervical lymphadenectomy in levels III, IV, VI bilaterally, and V on the left. The biopsy specimen revealed that a high percentage of the excised nodes were metastatic. The subsequent ultrasound confirms the presence of residual metastatic lymph nodes, leading to a surgical re-intervention for a modified radical cervical lymphadenectomy in stations II-V, followed by radioactive iodine therapy to eliminate residual thyroid tissue.

Discussions
Over the past decades, lymph node dissection for papillary thyroid cancer has been a subject of research, remaining controversial. Despite this, various scientific societies emphasize that, while not linked to increased morbidity in expert hands, it should be selectively indicated.

Conclusions
This case report underscores the significance of timely intervention and thorough management in metastatic papillary thyroid cancer. After surgery, the use of radioactive iodine proves crucial in addressing residual tissue. Multidisciplinary strategies, combining surgery and targeted therapies, showcase evolving approaches for effective treatment.

Keywords: papillary thyroid cancer, lymph nodes, management, Head and Neck, radioactive iodine

Complexity and Complications in Surgical Interventions: A Case Study Analysis

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Introduction
Hemicolectomy, a common surgical procedure, involves the removal of part of the colon for various conditions such as tumors or inflammatory bowel disease. Despite its advantages, hemicolectomy can be associated with risks and complications.

Case report
A 73-year-old male patient was admitted to the emergency unit with abdominal pain, vomiting and stoppage of bowel, diagnosed with subocclusive syndrome. Medical history shows type II diabetes. Further investigations, abdominal radiography and abdomino-pelvic CT, revealed the presence of a stenotic tumor in the transverse colon, for which it was decided to perform a right hemicolectomy. Following the forensic necropsy report, external examination of the corpse revealed signs of surgery, a xipho-suprapubic incision, an ileostomy hole and a drainage, through which blackish-red blood was draining. On internal examination, multiple lesions were identified. On examination of the head, the surface of the brain was found to be pale, and on the chest, a fracture of the left VI rib . In the abdomen, the peritoneal cavity contained about 700-800 ml of red-black blood and continent sutures in the remaining intestine and upper mesenteric vein.

Discussions
During hemicolectomy surgery, difficulties were encountered due to peritumoral inflammation, which led to avulsion and laceration of the middle colic vein. Hemostasis was obtained with difficulty by lateral venorrhaphy at the superior mesenteric vein over a length of 3 cm. The patient presented hemodynamic instability, following which he developed asystole and cardiopulmonary arrest. Death was due to hemorrhagic shock following a superior mesenteric vein continuity solution, confirmed by histopathological diagnosis, along with central nervous tissue with ischemic changes and anemia, and anemic-looking kidneys, all following hemorrhagic.

Conclusions
Hemicolectomies are complex surgeries and managing complications is crucial to the success of the procedure. Appropriate diagnosis and treatment requires a thorough preoperative assessment and careful attention to clinical detail to ensure an optimal outcome.

Keywords: hemicolectomy, hemorrhagic shock, surgical complications, vascular injury

Atypical case of pituitary aspergilloma transsphenoidally removed

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Introduction
The case presented is an aspergilloma found in the patient's sella turcica. The single mass was excided through FESS. The iatrogenic fistula was then obturated. Recovery time and regaining of quality of life were optimal.

Case report
A 53 years old female presented with migraine, nausea, vomiting, scotomas, left temporal hemianopsia and anterograde amnesia. Her symptomatology appeared 6 months prior, worsening and not responding to medical treatment.

Upon ENT clinical examination: symmetric facies, no adenopathy, normal buccopharyngoscopy, free nasal vestibulum. The nasal fossa presented a normal appearance, absent CSF leakage.

The paraclinical examinations highlighted a platelet distribution width of 15,9 fL.

A FESS surgical procedure was performed, including: a left uncinectomy, an antero-posterior left ethmoidectomy and a left sphenoidotomy. Opened the dura mater and penetrated the pituitary fossa, a soft, dark and necrotic mass was excided. Fat harvested from the left iliac fossa obturated the dural fistula, together with Surgicel. Finger gloves were inserted.

Amoxiplus (1000mg/200mg) and sea buckthorn oil were administered; the response was positive. The suturing stitches were removed seven days after with no complications. The symptomatology ceased entirely upon the removal of the mass.

Discussions
The peculiarity of the case lies in the location of the mass and in the approach chosen. Main surgical concerns were the incomplete obturation of the CSF fistula, uncontrolled bleeding, septal perforation, lamina papyracea injury, damage to crucial vessels.

Seen the circumstances, FESS was chosen due to its targeted approach and the minimally invasive nature.

Conclusion
To conclude, the case presented bears significant literary weight, seen the few similar ones already documented. The insurgence of the event in an immunocompetent patient, together with the complete disappearance of the whole symptomatology postoperatively, was remarkable. It is clear how vital it is to include a pituitary aspergilloma in the differential diagnosis when a pituitary mass is identified.

Keywords: Aspergilloma, FESS surgery, Iatrogenic CSF fistula

The multidisciplinary challenges of Morris Syndrome- a disorder grovelling on apparently normal infant females

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Introduction

Androgen insensitivity syndrome (AIS) is acknowledged as a rare, inherited, X-linked recessive disorder, leading to the most recurrent male pseudohermaphroditism. The coexistence of a male karyotype with a normal female morphology, marked as Morris syndrome, is followed in up to 40% cases by inguinal hernias containing a testis, typically passing unnoticed on seemingly normal infant females. This case report emphasizes the significance of coordinated multidisciplinary care in achieving favourable outcomes for patients facing this complex phenotypic anomaly.

Case report

An 11 year old girl presented to the ER with a progressive groin pain and weakness in the abdominal area. After the physical examination and CT scan, the patient was admitted for an emergency surgery due to bilateral incarcerated hernia. During the surgical procedure, after locating and separating the hernia from the adjacent tissues, the presence of a mass that seemed to be a testis was noticed. An excisional biopsy was performed and the resected pieces were sent for a histopathology exam, highlighting the existence of testicular parenchyma. Further genetic tests were performed such as chromosomal analysis showing a 46XY karyotype and check-ups of testosterone, luteinizing hormone and follicle-stimulating hormone, all of which confirmed the diagnosis of Morris syndrome. The patient was referred for estrogen replacement therapy and psychological consultation.

Discussions

Successful outcomes in such cases rely on vigilant intraoperative care, and meticulous postoperative monitoring. The occurrence of bilateral inguinal hernias in infant girls should accentuate the possibility of AIS, early diagnosis and hormone supplementation being vital in structuring the feminine phenotype.

Conclusion

Morris syndrome implies an effective multilateral management, the golden standard for a fortunate outcome, as demonstrated in this case report, being a definitive diagnosis which relies on the histopathological examination and further genetic tests.

Keywords: androgen insensitivity syndrome, Morris syndrome, multidisciplinary approach

Occult breast cancer treatment for an elderly patient: a challenge for the oncological field

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Introduction
Occult breast cancer (OBC) is defined as the presence of axillary lymph nodes metastasis in the absence of a primary mammary neoplasm at the preliminary assessment. It totals for 0.1-0.8% of all breast cancers worldwide and currently, there are no studies that specifically evaluate its etiology, making OBC occurrence a medical mystery, especially due to the lack of results during mammography screenings. This case report aims to evaluate a patient's journey dealing with OBC and describe the best course of treatment.

Case report
A 67 year-old woman presents at the Regional Institute of Oncology Iași for a routine mammography scan, uncovering "C" mammary density (ACR-BIRADS score), right breast asymmetry and right axillary adenopathy. An echography suggested 2 malignant axillary lymph nodes (15/26,5 mm), along with a Core-Biopsy of the right axillary region. The specimen concluded the diagnosis of stage III invasive ductal carcinoma (IDC), presenting tumor markers: ER and PR in >90% of the studied cells (along with Ki-67: 10%) while HER2 was negative. After 6 months of neoadjuvant hormonal-therapy, the patient undergoes a right Madden modified radical mastectomy with subscapular pedicle resection. Post-surgery, the studied specimen (resected breast, retropectoral and axillary lymph node tissue) discovered one metastatic adenopathy cluster and 21 axillary lymph nodes, 19 of them showing metastasis of IDC and GATA3 marker involvement. Following, the wound healed properly and she was discharged with a drainage tube, planned to be removed once drainage falls below 30 ml/day, marking an overall favorable outcome.

Discussions
Being rare and aggressive, the gold standard when dealing with OCD remains a modified radical mastectomy, chemo and hormonal therapy to reduce the risk of recurrence and further metastasis.

Conclusions
Everyday, research programs are keen to determine the etiology of OBC and understand this oncological mystery, offering the chance for earlier detection and better recovery rates.

Keywords: Breast cancer; occult; mastectomy; adenopathy

The successful story of a life-threatening hemorrhage that was solved with an emergency cephalic duodenopancreatectomy

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Introduction
The Wipple procedure, also known as duodenopancreatectomy, is considered one of the most complex surgeries and is commonly utilized for the treatment of pancreatic cephalic neoplasms, distal choledoc neoplasms, malignant duodenal tumors, and in some cases**,** chronic pseudotumoral cephalic pancreatitis.
The procedure involves removal of the antrum of the stomach, the first and second portions of the duodenum, the head of the pancreas, the common bile duct while blood supply to the liver is left intact, and the gallbladder.

Case report
A 60-year-old woman presented to the emergency unit in hypovolemic shock, low blood pressure, tachycardia and melena with fresh blood clots.
Pre-operatively, a superior digestive endoscopy was performed**,** which showed the existence of an ulcerated tumor with active bleeding, without the possibility of achieving endoscopic hemostasis.The patient was transferred to the imaging department, where the computed tomography revealed a retroperitoneal mass that was invading the duodenum.

Discussions
The computed tomography revealed the presence of an invasive retroperitoneal tumor in D2 with active intratumoral haemorrhage and intraluminal extravasation at level D2. Additionally, there were multiple connections between the tumor formation and the inferior vena cava, as well as between the tumor formation and the superior mesenteric package.
Intraoperatively, a large and very well vascularized gastrointestinal stromal tumor was found localized anterior by the inferior vena cava and the right side of the superior mesenteric pedicule, with multiple adhesions between it and the vessels and tumor.
It was performed an emergency cephalic duodenopancreatectomy with a hepatico-jejuno, pancreato-gastro, and precolic gastro-entero anastomosis.

Conclusions
Postoperatively, the patient presented an uneventful evolution and she was successfully discharged after 7 days. At the 6-month follow-up after surgery, the patient did not experience a tumor relapse.
The particularities of this case consist in the absence of complications in an elderly patient with a life-thtreatening hemorrhage from a duodenal gastrointestinal stromal tumor and in the complex surgical approach.

Keywords: hypovolemic shock, bleeding, cephalic duodenopancreatectomy, retroperitoneal tumour, GIST

Takotsubo Cardiomyopathy- Dying of a Broken Heart

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Introduction
Takotsubo cardiomyopathy, better known as broken heart syndrome, is a systolic disfunction of the apical segment of the left ventricle, mimicking a myocardial infarction. The shape the ventricle takes in diastole strongly resembles a Japanese octopus trap called "Tako-Tsubo", hence the eponymous syndrome. The purpose of this paper was to show the correlation between high-risk surgery and stress-induced cardiomyopathy.

Case report
A 63 year-old female presented to the emergency room complaining of dyspnea, dysphagia, accompanied by an anterior cervical mass. Blood tests showed hyperglycemia, uremia, and leukocytosis with normal levels of T3, T4 and calcitonin. CT result revealed a large inhomogeneous thyroid tumor of the left lobule compressing and deviating the trachea to the right, mediastinal lymphadenopathy and numerous pulmonary nodules bilaterally. Fine needle aspiration biopsy revealed an anaplastic thyroid carcinoma. At the time incipient diastolic disfunction was present. Surgical treatment consisted in left lobectomy of thyroid gland and left cervical lymphadenectomy. Postoperative, elevated levels of CK, CK-MB and troponin were noted. An emergency cardiology consult showed a dilatative cardiomyopathy of unknown origin, left bundle branch block, inferior STEMI and acute bilateral upper limb ischemia primarily on the right. Emergency coronarography was not performed and the patient was treated for myocardial infarction. What followed was hemodynamic instability and bradycardia, which eventually led to decease. Autopsy revealed cardiomegaly (300g), left ventricle hypertrophy and cerebral edema.

Discussions
Prompt diagnosis and treatment of Takotsubo syndrome, although difficult in this scenario, would have definitely increased the patient's survival chances. Classical clinical presentation of Takotsubo consists of: stressful trigger, chest pain, dyspnea, ECG repolarization changes (elevated ST, QT, low T wave, arrhythmias), cardiac enzyme rise, acute heart failure and serum catecholamines 30x normal.

Conclusions
Broken heart syndrome is an infrequent postoperative complication of stress-inducing surgeries. Physicians ought to be prepared to treat it nonetheless.

Keywords: Takotsubo syndrome, broken heart syndrome, anaplastic thyroid carcinoma

Management of T3 Laryngeal Squamous Cell Carcinoma

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Background
Head and neck squamous cell carcinoma is the seventh most common cancer worldwide, accounting for approximately 4,5% of all cancer diagnoses around the world, according to the 2020 GLOBOCAN estimations. Tobacco use, alcohol consumption and iradiation of the head and neck are amongst the leading contributors to the increasing HNSCC incidence.

Case presentation
A 49 year old male patient acusing symptoms of dyspnea and dysfagia presented into the ENT department. After computed tomography and tumour biopsy were performed, results came back positive for left hemilaryngeal squamous cell carcinoma T3N0M0, stage III. Patient was referred to the multidisciplinary care team for cancer patients, to establish the curative strategy. T3 laryngeal carcinoma is quite challenging to manage as the NCCN Guidelines leave options open in terms of treatment. Both conservative treatment and radical surgery are viable options. A common practice is to start with induction chemotherapy, assess the tumour's response, and then decide on the therapy. Medical practitioners have become more and more concerned with the quality of life the patient has after the surgical resection of laryngeal tumours. Keeping in mind the patient's age, the first line treatment was induction chemotherapy by docetaxel, cisplatin, fluorouracil (TFP) in four cycles. The tumour was unresponsive to ICT, thus, the decision of performing a total laryngectomy with complete cervical lymphadenectomy was made. Patient recovered well and was left with a permanent tracheostoma, leaving him with several option for voice rehabilitation such as esophageal speech, electrolarynx, or tracheoesophageal puncture.

Discussions
A considerable number of studies have shown that the social relationships of patients undergoing total laryngectomy suffer dramatically. A study of 150 patients from Babin et al. suggests that voice deprivation is a major social limiting factor and tends to push individuals into social withdrawal. In this context, larynx preservation treatment is preferred. Unfortunately, larynx preservation was not applicable in this case as the patient's tumour was unresponsive to TFP therapy.

Conclusion
Laryngeal squamous cell carcinoma can have devastating effects on quality of life as a consequence of treatment, which means voice rehabilitation is crucial for the patient's recovery.

Keywords: laryngeal cancer, squamous cell carcinoma, dysfagia

Ledderhose disease: from first symptoms to treatment

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Introduction

Plantar fibromatosis or Ledderhose disease, is a rare benign nodular hyperplasia of the plantar aponeurosis characterized by the growth of hard and round or flattened nodules on the soles of the feet. It typically affects both feet and progresses slowly. The nodules are often painless at first, but may cause pain when walking as they grow. People with Ledderhose disease may also have other conditions associated with the formation of excess fibrous connective tissue such as Dupuytren contracture. The exact cause of Ledderhose disease is not known, but heredity is thought to play a role in many cases.

Case report

We present the case of a 59-year-old male who presented to the Iasi ER, in 2023, with marked pain in both soles, difficulty walking, a sensation of tightness in the feet, discomfort when rolling the sole during walking, and the appearance of two nodules in the medial area of the plantar region, approximately 2 cm in dimension. Clinical examination revealed two round masses, well-defined, mobile with the underlying plane, without skin indentation.

Discussion

On ultrasound, the plantar fascia is bilaterally thickened from the normal 3 mm to over 10 and even 13 mm in the medial area of the right foot. No traumas, free fluid in deep layers, or other nodules are observed.
Sequential plantar fascia release was performed bilaterally in separate surgical interventions at three-month intervals, and total excision of bilateral nodules. The patient feels well postoperatively, having returned to normal activities within fourteen days.

There is no family history of Dupuytren's disease or Ledderhose. Possibly a carrier of the gene, but to confirm, genetic investigations are required.

Conclusion

Plantar fibromatosis is a rare benign aggressive disease which presents as nodules over the sole of the foot. The disease is usually manageable conservatively. A surgical excision of the nodule in severe cases will prevent recurrence.

Keywords: plantar fibromatosis, Ledderhose disease, hyperproliferation

Proximal humerus fracture and reverse shoulder arthroplasty

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Introduction
This case presents a complex proximal humerus fracture following a low-energy trauma treated by a total reverse shoulder arthroplasty with excellent outcomes.

Case report
61 year old female presented at the hospital following a fall. The physical examination highlighted a painful right shoulder with extensive bruising but no signs of vascular or nerve injury, nor of dislocation. These signs associated with crepitus suggested a closed fracture of the proximal humerus.

Xray and CT imaging revealed a closed, comminuted intraarticular fracture of the humeral head involving the displacement of the greater and the lesser tuberosities, anatomical neck and surgical neck, classifying it as a NEER 4 with a high risk of avascular necrosis of the humeral head.

The initial management included pain relief and joint stabilization through a splint before scheduling for a total reverse shoulder arthroplasty.

The outcome of the surgery, coupled with physiotherapy was excellent, with a ROM recovery of over 150° on shoulder flexion, 135° on abduction, over 40° on external rotation and complete internal rotation.

Discussion
In patients with no underlying arthropathies and around this age, an ORIF with plates and screws is usually preferred when a conservative treatment is not possible. However, hemiarthroplasty tends to be dependent on tuberosity healing, which in this case was shown to be unfavourable.

Conclusion
In this case, the choice of performing a TRSA allowed the patient to regain a very satisfactory ROM as well as quality of life while avoiding a prolonged convalescence and complications such as avascular necrosis of the humeral head.

Thus, the use of this technique shows a noticeable improvement in patient outcomes and may obtain more wide-spread indications in the future.

Keywords: Proximal humerus fracture, Total reverse shoulder arthroplasty

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