13th – 14th February
2020

OSCON

J.J.Strossmayer University of Osijek
Faculty of Medicine Osijek

Book of Abstracts

„MODERN-DAY GENETICS AND ITS FUTURE IN PERSONALIZED MEDICINE"

2nd International Translational Medicine Congress of Students and Young Physicians

Josip Juraj Strossmayer University of Osijek Faculty of Medicine

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OSCON 2020

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of Students and Young Physicians

„MODERN-DAY GENETICS AND ITS FUTURE IN PERSONALIZED MEDICINE"

13th – 14th FEBRUARY

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2nd INTERNATIONAL TRANSLATIONAL MEDICINE CONGRESS OF STUDENTS AND YOUNG PHYSICIANS

ABSTRACT BOOK

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2nd INTERNATIONAL TRANSLATIONAL MEDICINE CONGRESS OF STUDENTS AND YOUNG PHYSICIANS

CONTENT

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of Students and Young Physicians

WELCOME MESSAGE FROM THE PRESIDENT OF ORGANISING AND SCIENTIFIC COMMITTEE

13th – 14th February 2020

J.J.Strossmayer University of Osijek
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WELCOME MESSAGE FROM THE PRESIDENT OF THE ORGANISING COMMITTEE

Dear participants, lecturers, workshop leaders, and volunteers,

First, thank you for making OSCON 2020 possible. I would like to especially thank all the members of Organising and Scientific Committee for all the hard work they have put in this year's congress and for many tasks they have managed to successfully fulfil.

This year's main topic is *Modern-day genetics and its future in personalized medicine.* Why have we chosen this topic? This a field of medicine that has been mistakenly put aside for many years. That was because of a lack of understanding of the human genome and genetic models. Fortunately, new discoveries have put genetics in the spotlight as a target for new therapies, diagnostic tools, and personalized treatment.

Because of its relatively young origin, genetics is still controversial, and in some parts, we are still not sure where the limit for a human being to intervene is. As it is a very new, refreshing and "out of the box" approach to a patient, it is also a field in which we still don't have as much experience as we should.

That is why we are bringing experts to teach you how to think and how to use knowledge of genetics in differential diagnosis and therapy. You will all be proud members of a new era in medicine – personalised medicine. The key to the gate of personalised medicine is held by our knowledge of general genetics, pharmacogenetics, immunogenetics, oncogenetics, neurogenetics and all other parts of this science. Try to find your place in this future.

I am confident that you will enjoy this year's edition of OSCON, meet new friends, lose your possible fear of public speaking, and of course that you will learn something new.

I encourage you to ask questions and to "soak in" as much as you can.

Wish you all the best in your life and future career and be the best physician you can!

Luka Švitek
President of the Organising Committee

WELCOME MESSAGE FROM THE PRESIDENT OF THE SCIENTIFIC COMMITTEE

Dear Colleagues,

It is a great pleasure and honor to welcome you in the name of the Scientific Committee of OSCON.

Our main theme this year is reaching into every specialization in medicine. Genetics is such a broad field that not only plays a role in the diagnosis but also in the treatment of the patient. Besides caring for the patient, it opens the door for us to get a complete picture of the patient's family so that we could easily detect genetic diseases on time and take preventative measures according to them. This is also the definition of personalized medicine that we aspire to.

Our global theme is Translational Medicine which is based on close collaboration between fundamental and clinical research for faster and more effective implementation of discoveries in clinical practice. Considering genetics is the fastest-growing field of research and that it requires constant up-to-date, we found it ideally suited for our global theme

Scientific Committee strived to put together a quality program and to enable participants to hear directly from leading experts from the field of Genetics.

OSCON is a good opportunity for hearing outstanding domestic and international scholars and a great time for sharing ideas and experiences between students and graduates of biomedicine and healthcare. Our scientific program is designed to ensure that attendees gain a comprehensive understanding of the latest advances to later be able to improve their clinical skills.

Hopefully, throughout the next years our meeting will strengthen the relationship between basic and clinical sciences.

We are more than happy to welcome you to our faculty and we hope you will have a pleasant and useful time.

We are looking forward to meeting you at this Congress.

Nora Pušeljić
President of the Scientific Committee of the Congress

OSCON 2020

2 nd International Translational Medicine Congress
of Students and Young Physicians

ABOUT US

„MODERN-DAY GENETICS AND ITS FUTURE IN PERSONALIZED MEDICINE"

13th – 14th February 2020

J.J.Strossmayer University of Osijek
Faculty of Medicine Osijek

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GENERAL INFORMATION

Date: February 13 th (Thu) – February 14 th (Fri), 2020

Venue: University of Osijek, Faculty of Medicine, Josipa Huttlera 4

Main topic: „MODERN-DAY GENETICS AND ITS FUTURE IN PERSONALIZED MEDICINE"

Guest attendance policy: All registered participants are welcomed to all events and lectures. Wearing official conference badges is obligatory for entering any events.

Official language: English

Social media: oscon_mefos

OSCON

ABOUT FACULTY OF MEDICINE

The Faculty of Medicine of University of Osijek is the youngest medical faculty in Croatia. It was officially opened in 1968 as a branch of the School of Medicine in Zagreb, and in 2018 it celebrated 20 years as the independent medical faculty. The largest and main base of the faculty is the University Hospital Centre Osijek. The Faculty of Medicine in Osijek is the only medical faculty in Croatia that shares the same campus with its main teaching base, which is located within the University Hospital Centre complex.

Apart from a three-year undergraduate programme and two-year graduate study programmes (Biomedical-Laboratory Technologies) and a six-year long first degree study programme in Medicine, the Faculty of Medicine offers several postgraduate specialist study programmes and doctoral study programme. The aim is to provide students the highest level of knowledge and training adequate for the professions they are pursuing.

The Faculty of Medicine Osijek has defined main research areas and has been profiled by several interdisciplinary groups that link basic and clinical studies (translational medicine). We would also like to point out that, our two professors emeriti Antun Tucak and Savo Jovanović contributed to the faculty with their work and effort.

ABOUT UNIVERSITY OF OSIJEK

The Josip Juraj Strossmayer University of Osijek is a university located in Osijek, Croatia. It was founded in 1975 and it is organized in 12 faculties, 4 departments and one academy. University is a medium-size in comparison to other European Universities. University of Osijek has been developing into a modern European institution of higher education, and it is becoming a regional centre of knowledge, research and excellence. All efforts are directed towards the constant increase of teaching and studying quality. The University offers a high student standard concerning accommodation, learning facilities and other student services. The city of Osijek is known as the city of students.

ABOUT OSIJEK

Osijek is a modern Central European city with 17 city parks and gardens which make Osijek one of the greenest cities in Croatia. The City of Osijek is also famous for secession (a variation of art nouveau). The promenade along the Drava river is one of the longest walking trails in Croatia. Given the city of Osijek's long history, there's a variety of sights such as Tvrđa, a fortified part of the city from the 18th century.

Some of the most valuable examples of Baroque architecture in Croatia, such as the statue of Holy Trinity and General's-headquarters are located in Tvrđa and printed on 200 kuna bills. The tradition of higher education in Osijek exists since 1707 and today our university with its 17 faculties and departments is one of the most important scientific centres in Croatia. According to the latest official figures, University of Osijek has around 18 000 students enrolled in. Some of the notable people that lived in Osijek are two Nobel laureates in Chemistry. Lavoslav Ružička was awarded in 1939 and Vladimir Prelog in 1975. Both of them finished their secondary education in Osijek.

ABOUT CROATIA

Croatia is a Central European and Mediterranean country, bordering Slovenia in the west, Hungary in the north, Serbia in the east and Bosnia and Herzegovina in the south; the country also has a long maritime border with Italy in the Adriatic Sea. Croatia has an unusual shape (similar to a croissant) that is unlike any other country in the world, which is as a result of five centuries of expansion by the Ottoman (Turkish) empire towards Central Europe. It covers a land area of 56,691 square kilometres and has a population of about 4.29 million people.

The main population centres are Zagreb, the capital, Osijek in the northeast, the ports of Rijeka on the northern part of the coastline, and Split towards the south. Other well known towns include Dubrovnik, Makarska, Poreč, Rovinj, Opatija, Zadar and Šibenik.

On 25th June Croatia declared its independence from Yugoslavia and celebrated 27 years of independence in 2018 and, in that time, it has undergone many transitions – not least coping with the effects of the war in the early 1990s. It is a beautiful country and its unique scenery, culture, sights and beautiful coastline are certainly worth a visit. We've given you just a brief history of the country, but we hope you will experience it for yourself!

OSCON 2020

2 nd International Translational Medicine Congress
of Students and Young Physicians

LECTURERS

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13th – 14th February 2020

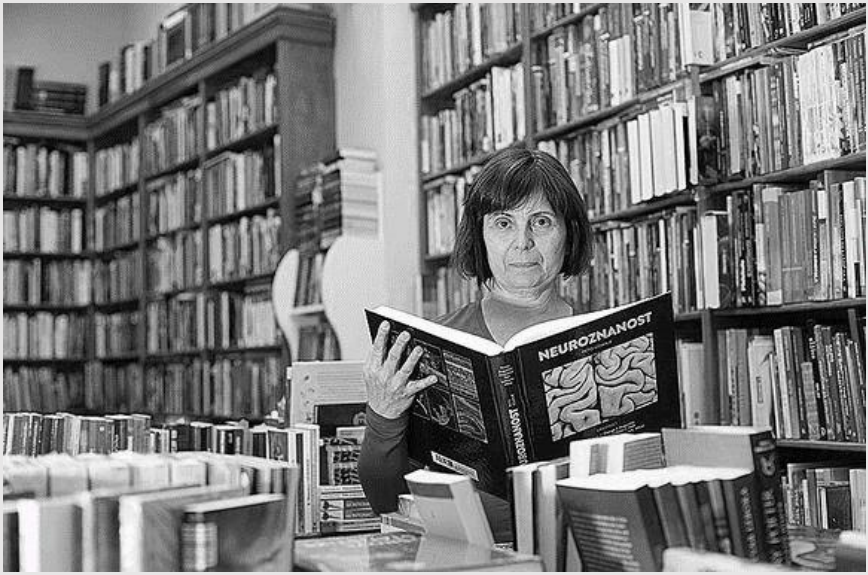
J.J.Strossmayer University of Osijek
Faculty of Medicine Osijek

Prof. Marija Heffer, MD PhD

Prof. Heffer graduated from School of Medicine, University of Zagreb in 1989 and did her postgraduate study in natural sciences. As a part of her scientific training, she studied biochemistry of glycolipids at The Institute for Zoology, Hohenheim University, Stuttgart and at The Institute for Cell Culture Technology, Technical Faculty, University of Bielefeld, Germany. Additionally, prof. Heffer was a guest scientist in the USA at Johns Hopkins School of Medicine and School of Medicine Yale. Since 2005 she has been appointed as a professor of Cell biology and Neuroscience at Faculty of Medicine, Josip Juraj Strossmayer University of Osijek.

Prof. Heffer is a neuroscientist with a very broad research area of interest, such as effects of stress on brain function, distribution of major gangliosides in various brain regions, roles of lipid rafts, dendritic morphology and spine density variations, genetic variations in circadian rhythm genes etc.

Regarding prof. Heffer's tremendous knowledge in many fields, such as biology, chemistry, neuroscience, genetics etc., we assure you that her lecture at OSCON will definitively be worth hearing and taking notes to!



Prof. Ivo Barić, MD, PhD

Prof. Barić has graduated at the School of Medicine, University of Zagreb. In 1992 he has further qualified as a pediatrician and obtained Ph.D. degree. He is a subspecialist in medical genetics, head of the Division for Metabolic Diseases, Department of Pediatrics, University Hospital Centre Zagreb, and also a professor of pediatrics at the University of Zagreb, School of Medicine.

Professor Barić has been a coordinator for Croatia of several scientific projects and published over 100 peer-reviewed publications. In one of them, previously unknown inherited deficiency of the S-adenosylhomocysteine hydrolase was described. For that work, he was awarded with "Horst Bickel-Award" and „Ante Šercer-Award".

Professor Barić is the current president of the Section for metabolic diseases and board member of the Croatian Paediatric Society. He is a full member of the Croatian Academy of Medical Sciences and a member of the Court of Honour of the Croatian Medical Chamber. He is head of the Committee for New-born Screening of the Ministry of Health, Republic of Croatia.

With his respectful scientific career in the field of medical genetics and metabolic diseases and broad clinical experience in metabolic diseases of children, you are guaranteed to acquire new, interesting and clinically useful knowledge.

Danijela Petković Ramadža, MD, PhD

Danijela Petković Ramadža has graduated from the School of Medicine Zagreb in 2004. In the following years, she was a research fellow and pediatric resident at the Department of Pediatrics, University Hospital Center Zagreb. Since February 2012 she is working as a pediatrician at the Metabolic Diseases Division. Currently, she is on a metabolic medicine subspecialty program.

Since January 2017, she is a teaching assistant at the Department of Pediatrics; Medical School Zagreb. She is actively involved in many national and international scientific congresses and symposia and is the author or co-author of several scientific papers mostly about rare genetic and metabolic diseases, so it is no surprise that she has also contributed to several European registries for rare metabolic diseases.

If you have registered to OSCON you will be able to enjoy yourself in lectures held by her and to learn a lot from broad clinical and scientific experience.

Asst. prof. Nina Pereza, MD, PhD

Asst. prof. Nina Pereza earned her medical and doctoral degree at the Faculty of Medicine, University of Rijeka, where she works at the Department of biology and medical genetics.
She is the co-author of over 20 scientific articles, which were published in various esteemed scientific journals related to reproductive biomedicine. Asst. prof. Pereza regularly participates at national and international congresses and is actively involved in different scientific projects. In addition, she is an editorial board member and reviewer in several medical journals. Her research interests are genetics and epigenetics of recurrent spontaneous abortion and spontaneous preterm birth.

Asst. prof. Silvija Pušeljić, MD, PhD

Asst. prof. Silvija Pušeljić graduated from the School of Medicine in Zagreb in 1994. She passed her specialty exam in pediatrics in 2001, and in 2002 she completed her postgraduate study in biomedical sciences. She has been working at the Clinical Hospital Center Osijek since 1994, and since 2016 has been the head of the Department of Neurology, Genetics, Metabolic Diseases, Endocrinology and Rheumatology in the same hospital.
She regularly participates in congresses in Croatia and abroad in the fields of genetics, metabolic diseases, and intensive care. Since 2001 she has been involved in teaching pediatrics at the School of Medicine in Osijek and in 2018 she was given the scientific title of senior research associate. In May 2013, she was given the professional title primarius and in April 2019 she has been recognized as a specialist in a narrower specialty - metabolic disease in pediatrics.
So far, she has mentored a total of 11 graduate papers, published 9 book chapters and published 37 scientific papers.
By attending Dr. Pušeljić's lecture on OSCON, we guarantee you many new interesting insights in the field of genetics.

Prof. Borut Peterlin,MD, PhD

Prof. Peterlin has been a Professor of Human Genetics at the Faculty of Medicine in Ljubljana since 2007 and is a Visiting Professor at the Universities of Belgrade, Tuzla, and Osijek. Prof. Peterlin earned his degree at the University of Ljubljana and Certificate in Human Genetics from Rene Descartes University Paris 1990. Prof. Peterlin is the head of the University's Clinical Institute of Medical Genetics in Ljubljana.

A major current professional and research interest involves the application of new genomic technologies in the diagnosis and identification of new mechanisms of human diseases. Professor Peterlin is an active member of the academic community, involved in many medical societies. He is currently a member of the Scientific Council for Neurogenetics of the European Association of Neurology and a member of the Working Group on Neurogenetics.

During his career, he distinguished himself as the coordinator of the GENEPARK project, led by a team of scientists who researched genomic biomarkers for Parkinson's disease. He has published over 200 papers indexed at Pubmed and he is a member of the editorial board in magazines: Public Health Genomics, Open Medicine and Balkan Journal of Medical Genetics.

OSCON 2020

2 nd International Translational Medicine Congress
of Students and Young Physicians

ePOSTER PRESENTATIONS

13th – 14th February 2020

J.J.Strossmayer University of Osijek
Faculty of Medicine Osijek

ePOSTER PRESENTATIONS

Zucić Josip → 16p13.11p11.2 duplication with epilepsy, cognitive deficit and phenotype dysmorphia – case report

Ilinca Dascalescu → A rare case of ascending aortic dissection complicated with ischemic stroke - A 22 year old patient with unknown bicuspid aortic valve

Toth Mate → "If there is no other way" – The limits of multivisceral resection in complicated colorectal cancer surgery

Burić Antonio → Acute intermittent porphyria and pregnancy – case report

Jovičić Miloš → An Uncommon Presentation of Temporal Lobe Epilepsy in a Young Adult

Edl Mia → Primary antiphospholipid syndrome: a case report

Zubak Hrvoje → Borderline cognitive ability with ADHD in XYY chromosomopathy: five case series

Jurić Ivana → Breathing disorders in Rett syndrome

Mariciuc Gabriel-Grigore → Surgical revascularization in axilar anevrism for polytraumatic patients

Lukić Matea → A Rare Double Aneuploidy Case: Down–Klinefelter

Smajić Matea → Sagliker syndrome – case report

Mariciuc Gabriel-Grigore → Surgical Revascularization in Chronic Mesenteric Ischemia

Dascalescu Ilinca → Challenges of diagnosis and treatment of a patient with resistant hypertension and bilateral renal artery stenosis

Gabunia Elene → Public Knowledge of Genetics in Georgian Population (2019 year)

Zagorac Irena → Cytodiagnostics of pancreatic neuroendocrine tumor: a case report

Getoš Josipa → Dilated cardiomyopathy: a case report

Laslo Dorian → From splenomegaly to Gaucher`s disease type I in an elderly patient: a case report

Perić Leon → Severe chronic asymptomatic hyponatremia: A case report

Vincetić Ivo → Coronary artery disease: A case report

Ormanac Klara → From abdominal pain to osteoporosis

Kljaić Lucija → Reccurent major depressive disorder with thyroiditis

Petrinović Matea → MODY – Maturity diabetes of the young

Raguž Petra → Mutation of GALT gene: Classic galactosemia – case report of 2 patients

Tomas Matej → The need for interdisciplinary collaboration during the treatment of orthodontic patients

Sabo Dea → Spondyloepiphyseal dysplasia congenita (SEDc) as a form of dwarfism

Šimić Ivana → HIV – positive young male patient with bipolar disorder and psychoactive substances addiction

Olujić Marija → Patient with newly diagnosed celiac disease and ataxia – case report

Neagu Ana-Maria → Congenital mesoblastic nephroma with uncommon evolution

Neagu Ana-Maria → Pulmonary thromboembolism associated with neoplasms

ePOSTER PRESENTATIONS

Pancu Cristiana Diana → The atrial myxoma and pseudovasculitis syndrome

Marušić Romana → Pheochromocytoma: a case report

Pintilii Oana Stefania → Multimodality imaging in cardiomyopathies: friend or foe?

Pušeljić Nika → Facial dysmorphism and intellectual disability in a rare 19q13.43 microdeletion syndrome

Štajdohar Kristina → Risk Factors for Laryngeal Cancer

Pintilii Oana → Simultaneous floating thrombus in the ascending aorta and pulmonary embolism

Vučić Domagoj → Takotsubo cardiomyopathy presenting with ST elevation in patient with an end stage renal disease on hemodialysis program: case report

Pavlović Vedrana → The importance of accurate diagnosis in case of stress and urge incontinence

Majer Jurica → The role of heredity in Bipolar affective disorder

Dascalescu Ilinca → The unspoken need of blood products required in acute aortic dissection

Pintilii Oana → Transient ischemic attack - the first manifestation of a rare genetic disorder

Dujmović Marin → Translocation Patau syndrome with long survival and comorbidity

Todić Lucija → Triple X syndrome as a cause of cognitive impairment

Laslo Dorian → Unusual retinal hemorrhages as a first sign of chronic myeloid leukemia: a case report

Burić Marko → Very long-chain acyl-CoA dehydrogenase deficiency – case report

Anđić Marija → Review of importance specific biomarkers in laboratory medicine for patients who suffer from rheumatic arthritis

Dozet Matea → Perioperative managment of patients receiving direct oral anticoagulants in dentoalveolar surgery

Jirouš Maja → Cancer organoids as preclinical model for drug development

Medač Petra → Review of recent advances in preimplantation genetic diagnosis

Veselski Karolina → A Review on Platelet Activating Factor Inhibitor Lexipafant: Could Lexipafant be a Standard in the Treatment of Sepsis?

16p13.11p11.2 duplication with epilepsy, cognitive deficit and phenotype dysmorphia – case report

Josip Zucić[1]; Antonio Burić[1]; Ante Listeš[1]; Silvija Pušeljić[1,2]

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2-University Hospital Center Osijek, Department of Pediatrics, Division of Neuropediatrics, genetics, metabolic diseases and endocrinology, Osijek, Croatia

Introduction: 16p13.11p11.2 duplication is a rare chromosomopathy, inherited in autosomally-dominant manner or sporadical, resulting in facial, limb and chest deformities, cognitive deficit, behavioral disorders such as ADHD or autism, epilepsy, cerebral and cardiovascular anomalies.

Case report: We present an 11 year and 8 month old female, a third child from two healthy, non-consanguine parents in their 30s. One case of chromosomopathy was found in the 4th branch of the family. Older brother(16) was treated for non-Hodgkin lymphoma. The patient had a globally retarded psychomotoric and a severely delayed speech development, thus classified as mildly mentally retarded. Phenotype dysmorphia includes triangular head shape, otapostasis, irregular dentition, gothic palate, epicanthus, bilateral clynodactyly and four finger furrow. She had a first seizure at the age of 5, and has since been well controlled on anticonvulsive therapy. MRI scan indicates corpus callosum hypoplasia. Karyotypization of the girl and her parents was performed at the Medical faculty in Osijek, showing excessive chromosome material on chromosome 16 and normal parental genotype, with cCGH showing the 16p13.11p11.2 duplication. Comorbidities include allergic asthma, recurrent pneumonia, autoimmune thrombocytopenia, neutropenia, Hashimoto thyroiditis and the left peroneal nerve palsy.

Conclusion: 16p13.11p11.2 duplication has a complex clinical presentation, linked to overall 25 genes. Duplication of 8 genes in 16p13.11 is linked to microcephaly, mental retardation, autism, ADHD, cerebral anomalies and aortal anneurism or dissection in early childhood, whereas the duplication of 17 genes in 16p11.2 is linked to macrocephaly, epilepsy, anorexia, depression, schyzophrenia and congenital cardiovascular anomalies. Our patient additionally has haematological and autoimmune manifestations rarely linked to this duplication.

Keywords: 16p13.11p11.2 duplication, phenotype dysmorphia, epilepsy, comorbidities

A rare case of ascending aortic dissection complicated with ischemic stroke - A 22 year old patient with unknown bicuspid aortic valve

Ilinca Dascalescu [1], Ana Maria Neagu [1], Oana Stefania Pintilii[1], Cristiana Diana Pancu[1], Gabriel Mariciuc[1], Asst. Prof. Carmen Elena Plesoianu[1,2], Dr. Dascalescu Daniel[2]

1-Faculty of Medicine, University of Medicine and Pharmacy "Grigore T. Popa", Iasi, 700115, Romania

2- Institute of Cardiovascular Diseases " Profesor Doctor George I.M. Georgescu", Iasi, 700503, Romania

Introduction: The risk of acute aortic events in patients with bicuspid aortic valve (BAV) disease is a controversial issue, the real risk being yet unknown.

Case study: We studied the case of a 22-year-old patient, with no known pathologies, sent from the department of neurology to our institute with stroke, left hemiplegia and severe chest pain. A thoracic CT-scan showed type A aortic dissection, with the fold of dissection stretching to the level of the right internal carotid artery, compressing it and thus causing neurological manifestations. At the transthoracic ultrasound, BAV was revealed. The patient did not receive fibrinolytic therapy, which made the emergency surgery possible to be performed under heparin therapy. Long term evolution was marked by amelioration of the motor deficit through recovery therapy. The young age, the underlying BAV and the symptoms of the patient fall into a minority of pathologies, making the diagnosis a challenge. Also, stroke as a complication to aortic dissection is represented by a small percentage, and performing cardiac surgery on a patient with stroke under heparin therapy increases the risk of hemorrhage. Moreover, if the patient would have received fibrinolytic therapy for stroke, emergency surgery for aortic dissection would have become an absolute contraindication.

Conclusion: The particularity of this case resides in the combination of a few rare factors: the very young age of the patient, the undocumented bicuspid aortic valve that precipitated the aortic dissection and stroke, as an uncommon complication to it. Knowing the correct management of this tight category of patients makes the difference. Moreover, an early echocardiographic evaluation should be available and mandatory at populational scale, making the occurrence of these events decrease dramatically through regular observation.

Keywords: aortic dissection, stroke, heparin

“If there is no other way” – The limits of multivisceral resection in complicated colorectal cancer surgery

Mate Toth [1]; Kornel Kovach MD [1]; Zsolt Simonka MD PhD [1]

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Introduction: Colorectal cancer (CRC) is the third most common cancer worldwide and it takes second place in tumor mortality. In the treatment of CRC, the leading role remains in surgery. According to recommendations for CRC surgery, the adequate intervention requires to be radical to achieve complete tumor removal with clear margins (R0), so the best possibility of long-term survival can be provided. However, this operative consideration may be difficult in cases, where CRC forms local adherence to or invasion of several, adjacent organs, so multivisceral resection (MVR) would be needed. But when should MVR be performed and what could be the limits of it?

Case study: 48-year-old male patient presented with the diagnosis of duplex colorectal cancer in the transversal and rectosigmoid segments. Explorative laparotomy revealed the local propagation of cancer and its critically borderline state of inoperability. Oncotherapy was started, but the patient interrupted the treatment after the third circle by his own free will and ignored it for several months. Since the further propagation of the CRC the patient presented again and accepted the modified oncotherapy. However, this form of the therapy resulted in the lack of success, which made the further therapeutic step questionable. After careful consideration MVR was decided as an optimal intervention. At last a great surgeon assumed to perform the MVR, which succeeded beyond the expectations and had a significantly positive effect on the patient’s survival.

Conclusion: MVR is a radical surgical operation, which aims the en bloc removal of cancer invading multiple adjacent organs. Despite of its radicality it is reasonable to push the limits of MVR in the treatment of CRC. As it is demonstrated in this study, MVR can lead to satisfactory results even in cases with complicating factors such as failure of the neoadjuvant oncotherapy or bad patient compliance.

Keywords: cancer, surgery, multivisceral resection

Acute intermittent porphyria and pregnancy – case report

Antonio Burić[1], Marko Burić[1], Matea Lukić[1], Livija Sušić[1,2]

1 – Faculty of Medicine, J.J. Strossmayer University of Osijek, Osijek, Croatia

2 – University Hospital Center Osijek, Croatia

Introduction: Acute intermittent porphyria (AIP) is a rare autosomal dominant disease caused by mutation in the genes associated with deficiency of enzymes needed for heme production. Episodes of acute porphyria can cause abdominal pain, vomiting, constipation, and diarrhea. A patient may also expirience muscle weakness, seizures, fever, and mental changes such as anxiety and hallucinations. In pregnancy, 54% of patients have an exacerbation of attacks in the form of seizures and abdominal pain which occur due to hormonal changes.

Case report: We present a case of a 46-year-old female who was treated for sterility and therefore, together with her husband, was undertaken karyotypization which turned out to be normal. At the age 27, she was treated at the Department of Gynecology for miscarriage in the first quarter, after which she was displaced to the Internal Clinic Department due to prolonged high intensity abdominal spasms, mostly in the epigastrium and under the right rib arch, followed by nausea, frequent vomiting and severe general poor condition. Based on the following laboratory findings : d-ALA 1135.6 µmol/dU, coproporphyrins 229 nmol/dU, PBG 352.8 µmol/dU, uroporphyrins 121 nmol/dU, the diagnosis of acute intermittent porphyria was confirmed. At the age of 33, she had a second miscarriage. A year later, in her third pregnancy, her disease was activated in a milder form due to renal colic and was therefore hospitalized to the Urology Department. She brought the pregnancy to the end and the baby was born by caesarean section at the 37th week of pregnancy. At the age of 38, she gave birth to her 2nd child. During pregnancy she was again monitored due to an increase in urine porphyrin, but without any acute signs of the disease. After that, the disease has been controlled by a proper carbohydrate-rich diet, adequate fluid intake, and avoidance of stressful situations and unsafe medications that can trigger attacks.

Conclusion: Although the prevalence of acute intermittent porphyria is 1-2/200 000 in general population, it should be suspected in women of reproductive age with prolonged abdominal pain of unknown etiology. A diagnosis is confirmed based on biochemical tests for increased urinary porphyrin concentration. At the stage outside the acute attack, the concentration of porphyrin in the urine may be normal, therefore it is of great importance to carry out genetic analysis of children whose parent suffers from acute intermittent porphyria, in order to make a timely diagnosis and to avoid triggers that can activate an acute, life-threatening attack.

Keywords: porphyria, pregnancy, acute abdomen

An Uncommon Presentation of Temporal Lobe Epilepsy in a Young Adult

Miloš Jovičić[1]; Jelena Šarić Sučić[1,2]; Stjepan Jurić[1,2]

1 – Faculty of Medicine, J. J. Strossmayer University of Osijek, Osijek, Croatia

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Introduction: Temporal lobe epilepsy (TLE) is the most common form of focal epilepsy. This neurological disorder is characterised by recurring focal seizures that originate in the temporal lobe of the brain. TLE can manifest with focal aware seizures, where the level of consciousness isn't altered, and focal impaired awareness seizures which impair consciousness to some extent. Even though the seizures start locally, in some cases they may spread to other parts of the cortex, presenting with unusual symptoms.

Case report: We present a case of temporal epilepsy in a 21-year old woman with complaints of unusual sensory visual phenomena. One month prior to her examination, she experienced a sensation of dizziness and swaying, after which she started sweating and vision in both of her eyes first turned yellow, then green, and finally black. According to her friend, both of her pupils were dilated. These symptoms would last approximately ten minutes. Afterwards, her vision would become blurred, and she would experience light flashes. These events would start with rising epigastric sensations. Later she remembered experiencing similar symptoms in the fourth grade of elementary school. The physical exam was normal, as well as the EEG. The laboratory tests showed her D-dimers to be elevated. Molecular testing discovered a gene mutation for PAI-1 (4G/4G), indicating she was homozygous for thrombophilia. After that, the patient was recommended to stop with the oral contraceptives she'd been taking. The patient's CT and CT venography showed no abnormalities and the MRI could not be performed due to the patient's permanent retainer. The EEG was conducted again after sleep deprivation, and it showed changes with paroxysmal tendency on the left side. The patient was discharged with Levetiracetam 2x500 mg, after which her condition showed significant improvement. The patient had reported experiencing three similar episodes afterwards, though shorter in duration. Her medication dose was elevated to 2x750 mg, after which she hadn't reported any returning symptoms.

Conclusion: This case demonstrates the involvement of the visual cortex positioned in the occipital lobe, which resulted in changes to the patient's vision. EEG after sleep deprivation showed typical spike-and-wave complexes on the left side. The PAI-1 (4G/4G) mutation incited us to do a CT venography, due to the possibility of dural sinus thrombosis. Even though nothing was found, the patient was advised to stop with the oral contraceptives, as to prevent future thrombosis. In the end, the patient responded well to the antiepileptic therapy.

Keywords: temporal lobe epilepsy, uncommon, visual cortex

Primary antiphospholipid syndrome: a case report

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Introduction: Antiphospholipid syndrome (APS) is an autoimmune multisystem disorder characterized by the clinical evidence of arterial or venous thrombosis, pregnancy pathology and/ or thrombocytopenia as a result of antiphospholipid antibodies (APL), a heterogeneous group of antibodies directed against phospholipid-binding proteins. APS is either primary or secondary to an underlying condition, commonly systemic lupus erythematosus (SLE). It is often presented with hypertension, deep vein thrombosis, stroke, livedo reticularis, miscarriages as well as neurological and cardiovascular symptoms.

Case report: This case describes a 37-year old male patient who is a heterozygote for factor II mutation. He suffered from multiple deep vein thromboses which subsequently resulted in gangrene and amputation of the third and fourth finger of the right hand. Considering earlier thromboembolic events and triple positivity of lupus anticoagulant (LAC), cardiolipin antibodies (IgG, IgM) and anti-beta2-glycoprotein I antibodies on two occasions in tests conducted more than 12 weeks apart, primary APS was diagnosed. He was treated with enoxaparin, oral anticoagulant (OAC), then new oral anticoagulant (NOAC) and acetylsalicylic acid. Symptoms and signs such as skin rash, skin lesions, Raynaud syndrome, arthritis, myalgia weren't present. The laboratory data revealed a presence of positive low-titre (1:100) antinuclear factor (ANF), positive anti-double stranded-DNA antibodies. Cardiolipin antibodies IgG and IgM were positive. Lupus anticoagulant test couldn't be performed due to anticoagulant therapy and therefore possible false positive results. NOAC therapy was discontinued because of the tendency to increase risk of development of arterial thrombosis in APS patients. Considering the patient's disease activity, he was also treated with prednisone and chloroquine in addition to vitamin D supplement. During the follow-up observation, positive ANF in low titre is still present, as well as low C3 and C4 levels, cardiolipin antibodies remain positive. The patient has no accompanying symptoms for systemic lupus erythematosus and laboratory results have suggested an active primary APS with hypocomplementemia. Therefore, additional immunosuppressive treatment with rituximab, monoclonal anti-CD20 antibody is considered.

Conclusion: It is important to diagnose APS as soon as possible because during its course, it can severely damage multiple organs. Cases in which APS should be considered as a diagnose are younger patients with multiple thrombotic events of unknown origin.

Keywords: antiphospholipid syndrome, anticardiolipin antibody, venous thrombosis

Borderline cognitive ability with ADHD in XYY chromosomopathy: five case series

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Introduction: 47, XYY syndrome is known to include signs such as increased height, enlarged risk of delayed cognitive development. Those boys can suffer from hypotonia or prolonged development of motor functions. Sometimes there are other signs like asthma, motor tics, and seizures. These individuals more often suffer from ADHD, depression or autism spectre disorders. Special physical characteristic as macrocephaly, macrodontia, pes planus, hypertelorism, clinodactyly, scoliosis; can be present, but they are not imperative with this syndrome.

Case study:

Case 1: Patient M. K. was discovered chromosomopathy through prenatal diagnostics. His karyotype is 47, XYY. He was without symptoms until the age of 16 months when he was hospitalised in status epilepticus, at about the same time he was processed by a psychologist that concluded his under average early psychomotorical development. He was controlled at the age of 3 years and was still showing the same signs of delayed development.

Case 2: Patient K. L. is 15 years old and has been recognised as borderline IQ with marked cognitive impairment. Phenotype is with minor dysmorphia – mild otapostasis and gothic palate. Karyotype analysis has also shown 47, XYY.

Case 3: Patient L. I. has been instructed to us because the paediatrician has recognized signs of ADHD. He was born in week 34 through caesarean section as a second twin with intracranial haemorrhage and perinatal infection. He has demonstrated signs of aggression and early psychomotor growth prorogation. He was diagnosed as 47, XYY. In the last appointment, he was 16 years old showing signs of ADHD, schooling difficulties, dyslexia, dysgraphia with verbal and intellectual ability impaired. Aggression and sexual impulses seem to increase.

Case 4: Patient L. L. has been diagnosed with 47, XYY through amniocentesis. Now he is 14 years old and has developed ADHD, myopia and cognitive disabilities.

Case 5: Patient P. M. expressed IUGR that was discovered with ultrasonography in the 37th week of gestation. As an infant, he has developed bronchitis and ASD was diagnosed by a cardiologist. Phenotype contains specifics including hypertelorism, ear pendants, gothic pallet, wide placed mamillae, and hypospadias. He was diagnosed when he was 4,4 months old as a 47, XYY syndrome.

Conclusion: All our patients have shown some form of cognitive impairment with demonstrated phenotype abnormalities. ADHD was often expressed in these patients. With this case series, we want to stress the importance of diagnosing XYY disorder as early as possible, in order to give patients adequate care – physical and work therapy as well as medical and psychological care through childhood and life.

Keywords: XYY, ADHD, cognitive impairment

Breathing disorders in Rett syndrome

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Introduction: Rett syndrome is a rare non-inherited genetic neurodevelopmental disorder that primarily affects females (1 in 12,000). It is commonly divided into four stages:

Stage I (early onset): starts between 6 and 18 months of age. Signs and symptoms in this stage are subtle (less eye contact, lose interest in toys, delays in sitting or crawling).

Stage II (rapid deterioration): starting between 1 and 4 years of age. Symptoms of Rett syndrome occur, such as slowed head growth, abnormal hand movements, hyperventilating, screaming or crying for no apparent reason, problems with movement and coordination, and a loss of social interaction and communication.

Stage III (plateau): begins between the ages of 2 and 10 years. Seizures may begin in this stage and generally don't occur before the age of 2.

Stage IV (late motor deterioration): This stage begins after the age of 10 and can last for years or decades. It's marked by reduced mobility, muscle weakness, joint contractures and scoliosis.

Most cases are spontaneous and 99% of them are caused by a mutation in the MECP2 gene. There is no cure for Rett syndrome. Treatment is symptomatic.

Case report: We present a case of 21-year-old woman with Rett syndrome. She was born at the 38th week through caesarean delivery. Normal development was observed until the age of 18 months, following which development regressed. The patient had been previously examined by several doctors, MRI of the brain was normal. The clinical and paraclinical findings prompted suspicions of Rett syndrome, molecular genetic test was carried out and results showed mutation on MECP2, indicating positivity for Rett syndrome. Atonic seizures started at the age of 3 and despite treatment with various combinations of antiepileptic drugs, she continued to have seizures. At the age of 10, parents noted breathing disturbances during awake, characterized by hypoventilation, apnea, breath-holding spells, episodic hyperventilation and air swallowing, what can be misdiagnosed as epileptic seizures. Because of breathing disturbances are suspected, videopolysomnographic was done and results suggested the evidence of central apneas during awake (AHI 45.8/h, lowest SpO2 27%), and obstructive sleep apnea during sleep (AHI 22.8/h, lowest SpO2 67%). Treatment was attempted in patient with non-invasive mask ventilation, but due to non-co-operation, it was abandoned, and parents rejected tracheotomy as a possible treatment.

Conclusion: Breathing disturbances are a major diagnostic and therapeutic problem in Rett Syndrome, because they are more pronounced during wakefulness.

Keywords: Rett syndrome, breathing disorders, apnea

Surgical revascularization in axilar aneurism for polytraumatic patients

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Introduction: The incidence of car crashes is rising and the resulting thraumatic lesions are extremely severe. The pacients with arterial lessions need urgent surgical intervention for saving their lifes. These pacients have a long-term and difficult post operatory recovery, until their process of reintegration in society is complete.

Case study: This is the case of a 34 years old male involved in a car accident with multiple thraumatic lessions. When the emergency medical team (EMT) arrived, they diagnosed him with alcoholic halene, head injuries and functional impotence of the right superior limb with open wound on the forearm. At the hospital, we performed a computer tomography and an angiography. Those paraclinic investigations showed us a medium hematoma and an anevrism of the right axilar artery. He came immediatly to the Vascular Surgery Departement where the doctors performed a subclavian-axillary bypass with a vein graft. Secondary, a fascietomy was performed at the anterior forearm box level, which resulted in the reperfusion of the arterial flow to the anterior forearm box area. Because of the edema that appeared after the reperfusion, we had to wait until we could have sutured the wound. .

Conclusions: After performing the interventions on this patient, he regained normal arterial flow in his forearm, and also, slowly, the functionality of his right superior limb. He was able to move his shoulder after only 3 weeks post operatory. The recovery process in these type of pacients might be long-term, but we expect to have a full recovery in a short period of time in our patient with an easy process of social reintegration.

Keywords: axilar anevrism, subclavian-axillary bypass, fascietomy, reperfusion edema

A Rare Double Aneuploidy Case: Down–Klinefelter

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Introduction: Chromosome anomalies are important causes of congenital malformations and miscarriages. They can be numerical and structural. Down and Klinefelter syndromes belong to the group of numerical chromosomopathies. Down syndrome is a trisomy of the 21st chromosome, while Klinefelter's syndrome is a trisomy of the sex (X) chromosome.

Case report: A 6-year-old boy, the first child of two healthy, young, nonconsanguineous parents (28-year-old mother, 31-year-old father), underwent karyotyping immediately after birth due to suspected Down syndrome. He was diagnosed with 48, XXY, +21. Sy Down, Sy Klinefelter. A positive family history was confirmed on the father's side (sister's nephew). The characteristics of trisomy 21 are currently clinically and phenotypically dominant. Phenotypically, epicanthus and ocular hypertelorism are observed, mongoloidly laid eyes, ears lower laid, high arch of palate. Extremities are symmetrical, visible palmar crease of the left palm, incomplete on the right, short and broad fingers on both hands. Smaller penis, testicles in scrotum. Lung auscultatory - normal, heart activity - rhythmical, clear tones and audible systolic murmur 2/6 along the left sternal edge. Cardiac tests revealed VSD, which closed spontaneously over time, but ASD II remained persistent and hemodynamically insignificant. Ultrasound revealed second degree subependymal intraventricular hemorrhage on both sides. Hypocsic-ischaemic lesions were also present on the frontoparietal part of both sides of cerebrum. Thyroid hormones were normal and ophthamological findings were proper, as well. Patient develops and progresses very well within complex chromosomopathy.

Conclusion: Down's syndrome and Klinefelter's syndrome are the most common chromosomal abnormalities in humans. The coincidence rate of Down's syndrome and Klinefelter's syndrome in the same patient is 0.098 % in newborns, and it has been found to be dependent on parental age.Therefore, we emphasize the importance of conducting additional diagnostic tests, in order to determine correct diagnosis of the patient and to be able to prescribe proper treatment and medical care for a specific patient with more than only one syndrome at the time, what is commonly found.

Keywords: chromosomopathies, Down syndrome, Klinefelter syndrome

Sagliker syndrome – case report

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Introduction: Sagliker syndrome (SS) is a rare syndrome caused by long-term heavy secondary hyperparathyroidism (HPT) in end-stage renal disease (ESRD). The diagnosis is established clinically, based on the most common SS signs: maxillary and mandibular disfiguring bone changes (usually progressing), pigeon chest, teeth abnormalities, deformed fingertips, short stature, hearing loss and psychiatric disorders.

Case report: The 38-year-old man, currently of low height, paraplegic, pigeon and barrel chested, with elongated upper extremities and deformed fingers, mandibular and maxillar asymmetric deformities, teeth malformations and depression presented in 2000 with painful cramps. By that time his obstructive uropathy resulted in ESRD and since then he has been treated with chronic hemodialysis. Due to extremely high serum parathormone (PTH), resistant to pharmacological treatment, subtotal parathyroidectomy (PTx) was performed in 2007. The principle of surgical treatment of secondary HPT includes leaving a part of glandular tissue (partial PTx) in place, or autotransplanting a part ectopically, in order to produce some PTH needed for its physiological function. However, he underwent parathyroid resurgery in 2010 for persistently high PTH of more than 2500 pg/mL (upper normal limit 69 pg/mL). In 2012 neck ultrasound showed two suspected parathyroids and in the same year one of the two glands was surgically removed. Postoperatively, the expected decrease in calcium and PTH serum concentrations did not occur again. It was at ten years after the diagnosis of ESRD when the patient began to notice more pronounced skeletal deformities (upper, lower jaw, extremities, with deformities of fingers, kyphoscoliosis) along with depressive disorder. In 2018 transversal spontaneous fracture of proximal diaphysis of the right femur was radiologically confirmed as a consequence of demineralization of the skeleton, with complete absence of radiologic presentation of both sciatic bones and ankylosis of the right coxofemoral joint. His laboratory findings still show extremely high PTH, low calcium and high alkalic phosphatase (1994 pg/mL, 1.89 mmol/L, 837 U/L, resp.) despite continuous appropriate pharmacological treatment.

Conclusion: SS was firstly recognized by Sagliker in 2004 and it has not been described in Croatia to date. Pervasiveness and knowledge of the syndrome is extremely low, thus patients with SS signs could be treated in a wrong way. The most efficient way of treating SS is PTx. However, it can only stop progress of the disease, but cannot return skeletal deformities. Recombinant PTH availability could help in decision for total PTx for SS in the future.

Keywords: end-stage renal disease, secondary hyperparathyroidism, parathyroidectomy, Sagliker syndrome, skeletal deformities

Surgical Revascularization in Chronic Mesenteric Ischemia

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Introduction: Chronic mesenteric ischemia (CMI) is an important death causing disease either by entero-mesenteric infarction or starvation. Diagnosis is often delayed before sending the patient to the vascular surgeon. Atherosclerosis is the main cause of CMI. Arteriography is essential in diagnosing CMI and delineating the atherosclerotic lesions. The revascularization procedure consists in an aorto-mesenteric bypass reconstructing 1-3 visceral arteries. The paper refers to one case of CMI treated in the Vascular Surgery Department, "Sf.Spiridon" Hospital Iasi during 2017.

Case study: The patient had suggestive symptoms for mesenteric and aorto-iliac diseases.Angio-CT exploration revealed specific lesions both for aorto-iliac disease and stenotic or occlusive lesions in the celiac trunk and mesenteric arteries.The case benefited from aorto bifemural bypass using a synthetic graft associated with aorto-AMS bypass with reversed vein graft. Immediate and remote results were favorable, with remission of intestinal symptoms, weight gain. Bypass patency was followed-up by angio-CT and eco-Doppler.CMI is a diagnostic and therapeutic challenge. Open surgery provides symptom remission in 90% of cases. Permeability to 5 years is 80-90% for open surgery, higher than through endovascular therapy. Average permeability of the two types of intervention is 70% at 5 years, similar to the infraaortic bypasses.

Conclusions: The case was successful at the end of the procedure and over the two years follow up. The case came to the Vascular Surgery Department, "Sf.Spiridon" Hospital Iasi for symptoms of aorto- iliac disease and also chronic mesenteric ischemia. With an appropriate surgery both problems were solved and the patient was succesfully inserted in their familie and society.

Keywords: mesenteric bypass, chronic mesenteric ischemia, aorto-bifemoral bypass

Challenges of diagnosis and treatment of a patient with resistant hypertension and bilateral renal artery stenosis

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Introduction: Resistant hypertension is defined as blood pressure that remains high despite simultaneous use of three antihypertensive drugs, including a diuretic.

Case study: We studied the case of a 66-year-old patient who presents with angina and headache in the context of uncontrolled blood pressure values, in defiance of quadruple antihypertensive therapy. The physical examination revealed high blood pressure (BP = 220/110 mmHg), with a median BP of 176/88 mmHg at ambulatory blood pressure monitoring and severe scoliosis. The additional investigations showed left ventricular hypertrophy with preserved ejection fraction, hyperkalemia, normal lipids and carbohydrates values, proteinuria, chronic kidney disease and absence of inflammatory syndrome or anemia. In the context of multiple cardiovascular risk factors and angina, coronary angiography was performed, which excluded coronary stenosis. The onset of hypertension at an young age determined us to search for a secondary cause. There were no signs for Cushing's syndrome, bradycardia and the absence of palpitations excluded pheochromocytoma. Serum potassium was constantly high, so there was no primary hyperaldosteronism. The abdominal echography objectified small kidneys with irregular contour. Renal arteries angiography showed bilateral renal artery stenosis. Moreover, there were important limitations for both medical and interventional treatment due to hyperkalemia, bradycardia and severe anatomical particularities of the spine. The medical treatment was adjusted to a double dose of diuretic, increased dose of calcium blocker, attempting to reintroduce spironolactone along with the normalization of potassium values.

Conclusion: The particularity of this case resides in both the diagnostic and therapeutic challenges of a grade III secondary hypertension. Due to multiple associated pathologies, the medication and interventional solutions were limited, making us ask: in the absence of any interventional possibilities, what could be the solution for this hypertensive patient?

Keywords: secondary hypertension, angina, headache

Public Knowledge of Genetics in Georgian Population (2019 year)

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Introduction: In March-April 2019 we conducted a cross-sectional study with the aim of evaluating the knowledge of medical genetics in the lay Georgian population. Every person was eligible to participate. Topics included gene biology, genetic diseases, and genetic testing.

Materials and methods: Data was collected via scripted and confidential in person interviews. Interview questions were closed ended or multiple choice and were divided into five topics. Interviews were conducted with 415 participants, diverse in age, religious affiliation, education level, and profession. Most participants were 18-45 yrs. (71,6%), followed by 46-65 yrs. (22,1%), and >65 yrs. (6,3%). Most participants held an academic degree from a university (75,9%), while those remaining had only obtained a professional bachelor's diploma (12%) or only completed secondary education (12%). The majority identified as Orthodox Christian (86,8%), while small minorities identified as non-religious (9%), Muslim (2,4%), or other (1,5%). Of these, most considered their religious involvement somewhat active (45%) or passive (44.6%), with only a minority being active (9,9%).

Results: Topic 1- General knowledge about genes and diseases: There were 12 questions on this topic. The question which the most participants answered correctly (84,2) was, “is gene a disease?” The question which the most participants answered incorrectly (45,4%) was, “the genotype is not susceptible to human intervention.” Topic 2 – Disease-related concepts: There were 5 questions on this topic. The question which the most participants (91.3%) answered correctly was, “the onset of certain diseases is due to genes, environment and lifestyle”. Topic 3 - Facts related to cancer development: There were 6 questions on this topic. The majority (54.1%) of participants correctly knew that this statement is not true: genetic testing is only used to determinate whether you have cancer right now. 34.5% of participants were incorrectly informed that only women can have an altered breast cancer gene. Topic 4- Perceived Genetic Knowledge of Participants: There were 5 questions on this topic. 34% of participants were well informed about the possibility of early detection of certain disorders using DNA-testing. Topic 5 – Genetic Testing: There were 6 questions on this topic. 48.3% of participants thought they had adequate knowledge of their rights to refuse DNA testing (and this was highest result of „well know”)

Conclusion The results of this research serve to improve genetic knowledge depending on performing educational activities. This therefore will improve healthcare level.

Keywords: Genetics, Georgia

Cytodiagnostics of pancreatic neuroendocrine tumor: a case report

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Introduction: Pancreatic neuroendocrine tumors are rare tumors of the pancreas that arise from abnormal growth of pancreatic endocrine cells and account for only 1-2% of all pancreatic neoplasms. Fine needle aspiration of pancreatic material, performed during the endoscopic ultrasound procedure, is a fast and safe procedure, with a low range of complications that provides material for cytodiagnostics, and thus helps to determine the nature of pancreatic mass and, with enough material, can provide a specific diagnosis.

We report a case where cytology examination, provided with sufficient material, diagnosed rare tumor that was afterwards confirmed by histopathology.

Case study: A 42-year-old man underwent endoscopic ultrasound (EUS) examination after recurrent episodes of abdominal pain. The procedure revealed pancreatic masses that measured 21x26mm, and 30mm in diameter, as well as one enlarged lymph node, measuring 15mm in diameter. Fine needle aspiration of aforementioned masses was performed during EUS and material was sent for cytologic examination.

Cytological spreads revealed the same microscopic image in specimens acquired from each mass. It was the case of a relatively monomorphic population of round to polygonal cells (plasmacytoid like), with round to oval nucleus whose chromatin was finely dispersed ("pepper-salt" appearance). Cell cytoplasm was sharply marked with occasional fine reddish granules. The lining of specimens contained numerous individual cells, naked nuclei, clusters of finely ramified capillary networks and abundant blood cells. Considering the specific appearance and structure of cells, including reddish cytoplasmic granules, suspicion was raised that this was the case of neuroendocrine pancreatic tumor, so additional immunocytochemistry was performed. The remaining slide was stained for synaptophysin, a specific marker for neuroendocrine tumors. Analysis of the slide revealed diffuse positivity of tumor cells for synaptophysin, and diagnosis of neuroendocrine pancreatic tumor was made.

Conclusion: The patient underwent surgery for tumor excision, which was sent for patohistological examination. After the initial assessment of hemalaun-eosin stained slides, which showed a tumor consisting of mostly uniform cells arranged in a trabecular pattern with small nuclei, additional immunohistochemistry was performed, that included synaptophysin, chromogranin and CD56 markers. Positivity of tumor cells for those stains confirmed cytological diagnosis of neuroendocrine pancreatic tumor.

EUS-FNA in diagnosing pancreatic lesions is a highly specific and sensitive method, that can provide accurate diagnosis in a short period of time (a couple of days), and thus shorten the time for further patient treatment.

Keywords: cytodiagnostic, pancreatic neuroendocrine tumor

Dilated cardiomyopathy: a case report

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Introduction: Dilated cardiomyopathy (DCM) is a progressive disease of heart muscle characterized by dilation of all heart chambers, primary left ventricle and atrium, that is associated with systolic and diastolic dysfunction of left ventricle. DCM also can lead to heart valve problems, arrhythmias and blood clots in the heart. Around 30% of DCM has a genetic cause but it can also occur due to viral infections, alcoholism and other. DCM is common cause of heart failure.

Case report: A 25-year-old male was diagnosed with dilated cardiomyopathy four months ago and treated with heart failure therapy (ramipril, bisoprolol, furosemide, eplerenone). From the family anamnesis we can see that the patient's mother has an DCM that manifested at the age of 40. He presented with complaints of breathlessness on minimal exertion in past few days, dyspnea in rest last night and also not tolerating supine position. Therapy with furosemide and ramipril was discontinued by patient because of symptomatic hypotension. On examination he is conscious, tachydispnoic, with poor blood flow in skin and visible mucosa. Examination of chest showed crepitation in the basal parts of the lung and muffled heart tones. Liver was enlarged. Laboratory findings showed increased ALT, AST and GGT indicating liver failure and slightly reduced eGFR indicating kidney failure. ProBNP was markedly elevated because of heart failure. A scan of the thoracic organs showed a markedly enlarged shadow of the heart at the expense of all cavities and cranial redistribution of pulmonary blood flow. From electrocardiogram we see sinus tachycardia and left atrial dilatation. The echocardiogram showed that all four chambers of the heart are dilated. Systolic function of left ventricle is greatly reduced (EF 17%) as of the right ventricle. Diastolic dysfunction of left ventricle is III/IV degree. Aortic valve stenosis and regurgitation is absent. Mild mitral regurgitation is present. The maximum pressure in the right ventricle is 54 mmHg which indicates mild pulmonary hypertension. The patient's condition did not improve despite intensive therapy (dopamine, dobutamine) so he was moved to clinical hospital where an invasive cardiac processing (coronary angiography, catheterization of the right heart) was performed, after which the patient was placed on a transplant list and had heart transplantation 3 weeks later.

Conclusion: In this patient there was an early expression of DCM, which is probably genetically conditioned, followed by a progression that required cardiac transplantation as a form of treatment for the disease.

Keywords: dilated cardiomyopathy, heart transplantation

From splenomegaly to Gaucher`s disease type I in an elderly patient: a case report

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Introduction: Lysosomal storage diseases (LSDs) are hereditary diseases where mutation affects genes which encode lysosomal enzymes. In this group of diseases are classified sphingolipidoses, disorders where lysosomal enzymes necessary for degradation of cell membrane components, are not functional because of mutations in their genes. The most common type of sphingolipidosis is Gaucher`s disease (GD). GD is an autosomal recessive disease in which mutation affects the gene (GBA1 located on chromosome 1) responsible for glucocerebrosidase which hydrolyses glucosylceramide into glucose and ceramide. Decreased enzyme activity causes accumulation of glucosylceramide in lysosomes.

Case report: The patient was first examined in the haematological ambulance at the age of 75. Then she had normocytic anaemia (haemoglobin 108 g/L)and thrombocytopenia (thrombocytes 67x109/L). The patient had splenomegaly (palpable about 3 cm), hepatomegaly and gallstones. At that time, she did not agree to any diagnostic examination. In the age of 82 haematologist examined the patient again, than she had a significantly larger spleen 23x7, 6 cm, hepatomegaly and gallstones were still present. Furthermore, neurological examination was normal. Blood test results revealed even lower levels of haemoglobin (85 g/L) and thrombocytes (19X109/L). The patient was genetically tested for GD and the results revealed an elevated concentration of glucosylsphingosine (386,9 ng/ml) while β-glucocerebrosidase was 0.0 μmol/L . In the patients genotype two mutations were confirmed first of which was heterozygous recombinant allele derived from a recombination between the functional GBA gene and pseudogene GBAP1 and the second was missense mutation c.(1226A>G); (1265_1319del55; 1448T>C; 1483G>C; 1497G>C). Based on these results we diagnosed the patient with GD type I.

Conclusion: GD is rare in Croatian; diagnosis at older age is also relatively rare.GD should be considered a differential diagnosis when thrombocytopenia and splenomegaly are found in elderly patient. Although GD type 1 can be diagnosed at any age, it is rarely diagnosed at those ages (more than 80), however it is important to test patients in those ages for GD with the purpose to test progeny for GD.

Keywords: Gaucher, lysosomal storage disease, sphingolipidoses

Severe chronic asymptomatic hyponatremia: A case report

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Introduction: Hyponatremia is primarily a disorder of balance of water in the body. It is associated with absolute or relative excess of water in relation to overall sodium in the body.

Case report: Here, we report a case of a 51-year-old woman who was being treated for depression, hypothyroidism and arterial hypertension. During the preoperative preparation for umbilical hernia, laboratory tests showed sodium = 121 mmol/L and the patient was referred to the emergency room. The patient received 0,9% sodium chloride(NaCl) 500 mL. Concentration of sodium in serum afterwards was 122 mmol/L and the patient was reffered to an endocrinologist. While studying previous medical documentation, it was revealed that sodium values have been reduced over the last three years. The patient did not complain about any difficulties, and was of good clinical status. Laboratory tests once again confirmed hyponatremia and hypoosmolarity (125 mmol/L; 257 mOsm/kg). The basic laboratory findings, thyroid-stimulating hormone(TSH), free T4(fT4), cortisol and cortisol in the ACTH stimulation test were within reference values, as were the attached findings of cardiac and pulmonary RTG, mammography, abdominal ultrasound and gynecologist findings. The patient did not agree to change her therapy for depression, fluoxetine with flufenazine. The diagnosis given was severe chronic asymptomatic hyponatremia, with urine osmolality and sodium in urine (396 mOsm/kg; 80 mmol/L) which suggested syndrome of inappropriate antidiuretic hormone secretion (SIADH). After limiting the fluid to 800 mL there was an increase of sodium (128 mmol/l) and surgery was approved.

Conclusion: The usage of some antidepressants and antipsychotics can lead to SIADH and hyponatremia. In these patients, hypothyroidism and adrenal insufficiency (TSH and cortisol) should be ruled out and fluid intake should be limited to 500 mL less than daily diuresis or 800 mL throughout the day.

Keywords: Hyponatremia, hypothyroidism, depression, antidepressants, antipsychotics

Coronary artery disease: A case report

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Introduction: Coronary artery disease (CAD), also known as coronary heart disease (CHD) or ischemic heart disease (IHD), involves the reduction of blood flow to heart muscle due to build-up of plaque in the arteries of the heart. It is the most common of the cardiovascular diseases. Types include stable angina, unstable angina, myocardial infarction and sudden cardiac death.

Case report: We report a case of a 60-year-old woman, who was taken to the accident and emergency room. For two months she had been experiencing a severe chest pain during physical effort. In time, ailments progressed and eventually her symptoms started to occur even in contact with the cold air. The pain subsides when she rests. She did not have any respiratory problems nor any signs of dyspnea. She had multiple risk factors for coronary disease such as unregulated hypertension and hyperlipidemia, also her family's medical history was positive to cardiovascular diseases. Earlier this year she has developed hypertensive crisis that caused subarachnoid bleeding. In her reports, chest X-ray was clean and there were no signs of ischemia on ECG. Cardioselective enzymes were within reference interval, but because of her significant anamnesis we suspected coronary artery disease. Echocardiography has been made, which was clean with an estimated 64% ejection fraction. Coronarography showed significant stenosis of LAD. She got 2 stents and therapy for her coronary disease was prescribed.

Conclusion: Regardless of today's technology techniques which are very specific in discovery of coronary diseases, such as ECG, echocardiography, X-ray and laboratory reports, the rule of taking anamnesis is equally important in recognition of coronary disease and its future treatment.

Keywords: Coronary artery disease, coronarography, LAD

From abdominal pain to osteoporosis

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Introduction: Osteoprosis is a progressive, systemic metabolic disease characterised by reduced bone mass,bone architecture disorder and fragility of the skeleton. Estrogen inhibits cytokines that activate osteoclasts, and opposes the impact of parathyroid hormone, which initiates bone reabsortion and calcium mobilization. Lack of estrogen therefore, increases the risk of osteoporosis in women in postmenopause.

Case report: This case describes a 70-year-old female, that suffers from arterial hypertension, coronary disease and nefrolitiasis. In the last three months, she expressed a moderate pain in the lumbosacral part of the spine. She entered menopause at the age of 36. Her mother suffered from osteoporosis. When admitted to hospital, she felt a strong abdominal pain, as well as a mild pain in the lumbosacral part of the spine. Clinical examination excluded any cardiovascular or gastrointestinal disease. An X-ray was performed,and several fractures were identified: fractures of the vertebral body (L1-L3, Genant 2) and a fracture of the 11th thoracic vertebra (Genant 1). Using densitometry, osteoporosis of the spine and hip was verified (T-score: -3,3 and -2,9). Any secondary causes of osteoporosis were excluded. The rest of the clinical examination (including lab-tests,heart and lung X-ray, mammography,abdominal ultrasound and gynecological exam) showed no deviations. Interpretation of the clinical examination strongly indicated postmenopausal osteoporosis with multiple compressive fractures. Additional examination also identified damaged glucose tolerance. As for therapy, teriparatide (20 µg s.c. during 24 months) combined with colecalciferol was advised. She was mobile, but with help of analgesics and Jewett orthosis (antalgic gait).

Conclusion: Although the differential diagnostics of acute abdominal and thoracic pain includes a large spectre of disorders,in patients older than 65 years-of-age and ones that have a history of osteoporotic fractures in family, compressive fractures should also be considered. It is of essential value to timely diagnose osteoporosis,and with that, prevent any future compressive fractures, as it improves the life quality of the patient.

Keywords: thoracic pain, abdominal pain, osteoporosis, compressive fractures, teriparatide

Reccurent major depressive disorder with thyroiditis

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Introduction: Autoimmune thyroid diseases such as Graves' disease and Hashimoto's thyroiditis are defined by elevated serum levels of anti-thyroid antibodies and thyroid hormone dysfunction. Anti-thyroid antibodies could also affect extra-thyroidal tissues and organs. A higher lifetime prevalence of depression was found in TPOAb positive versus negative individuals. Thyroid autoantibody's pathophysiological effect in depression may be cytokine mediated. Higher proportions of Th17 cells are present in patients with Hashimoto's thyroiditis and Graves' disease and serum IL-17 levels are significantly increased in patients with intractable Graves' disease. Depression has been frequently associated with elevated levels of proinflammatory cytokines, especially IL-6 and tumor necrosis factor (TNF)-α.

Case report: A 49-year-old woman was admitted to psychiatry department after worsening of mental state. The patient is not married, has no children and unemployed. Patient's father had unknown psychiatric disease. On admission, patient revealed restlessness, moodlessness, depressed mood, impaired appetite, impaired sleep dynamics in the last five days. Hematologic and biochemical profiles including renal function tests and urine examination were all within normal ranges. EEG showed moderately dysrhythmic irritation. The patient has been hospitalized several times with recurrent major depressive disorder with psychotic elements and auditory hallucinations. The first hospitalization was when the patient was 24 years old. Psychopharmaceutical treatment has been administered which resulted with positive reaction and less productive symptomatology. Sociotherapeutical treatment has also been administered. The patient has a history of hypertension, gastritis and chronic thyroiditis. The patient has been complaining about pressure in chest, dry throat and shortness of breath. The patient has gained 12 kilograms. The thyroid gland is palpable and the hormones are currently within normal ranges.

Conclusion: The relationship between major depressive disorder and thyroid disorder has been established. It is important to know the family history and biochemical profiles to correctly diagnose the patient.

Keywords: major depressive disorder, autoimmune, thyroid

MODY – Maturity diabetes of the young

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Introduction: Diabetes is a group of metabolic diseased characterized by hypergycemia, resulting from defects in insulin secretion, insulin action, or both. Maturity-onset of diabetes of the young is defined as a form of monogenic diabetes, characterized by autosomal dominant inheritance and onset before 25 years of age. Mody usually occurs as a result of mution of genes encoding transcription factors important for the regulation of insulin gene expression and the expression of genes encoding proteins involved in glucose transport and metabolism in beta cells.

Case report: An asymptomatic patient at the age of 16, on several occasions measures random fasting glycemia in the range of 6.8 to 7.2 mmol/L. From her family's medical history, it was found that the patient's sister, mother and maternal grandparents had diabetes (3 generations). Her grandfather was diagnosed with diabetes at the age of 40, not treated. Mother's diabetes was diagnosed at the age of 24, as a part of preoperative treatment; she did not consent to the proposed insulin treatment. Diabetes mellitus was also diagnosed in her sister, at the age of 18 and she is currently on insulin therapy. Physical examination shows that she has a normal BMI, with no classic signs suggesting insulin resistance. In the lab. findings: plazma glucose level in OGTT with 75 g glucose and with insulin determination (0, 60, 120 min): 8.2 - 11.1 - 11.4 mmol/L, insulin 16.6 - 31.6 - 34, 5 mmol/L; HbA1c 6.9%; finding of specific autoantibodies (anti GAD, ICA, anti IA-2) negative. Genetic testing confirmed that the patient had a specific mutation of the c.683C> T GCK gene, or MODY type 2. Given this, insulin therapy was not required in patients, and only general treatment measures were recommended. GCK is a glucose sensor expressed in pancreatic β-cells. This mutation causes decreased glucose sensitivity due to phosphorylation defect. As a result, beta-cells are enable to responde appropriately to the degree of glycaemia.

Conclusion: Mody is reported to be the most common form of monogenic diabetes and affects 1-2% of all patients in Europe. 5% of individuals diagnosed with diabetes before age of 45 have MODY. 80% of those individuals are misdiagnosed as having type 1 or type 2 diabetes. It is important to recognize patients with MODY, because of the different approach in treatment. Dietary intervention alone is usually advised for these patients, as pharmacological intervention is not required to control hyperglycemia and prevent diabetic complications.

Keywords: MODY, GCK, monogenetic diabetes

Mutation of GALT gene: Classic galactosemia – case report of 2 patients

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Introduction: Galactosemia is a disorder that affects how the body processes a simple sugar called galactose. Classic galactosemia is the most common and severe form of the condition caused by a mutation in the GALT gene. Complications appear within a few days after birth and include feeding difficulties, jaundice, liver damage, sepsis and shock. There is also an increased risk of delayed development, cataract and intellectual disability.

Case report: This case represents two boys with classic galactosemia, both born healthy after a full-term pregnancy to two healthy nonconsanguineous parents. The patient, A.B., 20 days postpartum, was relocated from the local hospital due to relapsing vomiting and weight loss. In the family, 2 brothers on the maternal side died from jaundice at infantile age. 6 days after admission, the clinical symptoms include paralytic ileus, liver and kidney failure and a visible cataract. There's also a development of a septic shock as well as suppurative meningitis, caused by a resistant E.coli. Anamnestic data and clinical symptoms of the illness suspect galactosemia. Immediately, a lactose-free diet was included, as well as an antimicrobial and supportive therapy. The diagnosis was confirmed by finding increased lactose levels, both in serum and urine, and measuring reduced galactose-1-P-uridylyltransferase enzyme activity: 0.95U/ml (norm. 4-20). GALT gene analysis confirmed the mutation (p.Q188R/p.Q188R). Today, the boy is 12 years old, entirely abides the diet regime, but has a severe cognitive deficit with elements from the autistic spectre. Both eye cataracts were surgically taken care of, other symptoms of galactosemia mostly disappeared. The second patient B.S., develops indirect hyperbilirubinemia 48h postpartum with occasional vomiting, and at 4 days old, he shows signs of extreme jaundice and weight loss. Like the patient A.B., he develops sepsis caused by E.coli., and this awakens the suspect on galactosemia. He began with the same therapy as A.B. Serum galactose: 832 umol/L, urine galactose: 18,1 mmol/L, enzyme activity: 0,83. GALT gene analysis showed p.K285N mutation, which confirmed the diagnosis of classic galactosemia. B.S., today at the age of 7, is developing normally, without any signs of somatic or cognitive deficit.

Conclusion: Although it's a very rare genetic disorder, galactosemia should be considered. It's of extreme importance to consider galactosemia at the first signs of any symptoms, as the disorder is curable and doesn't have any repercussions if timely diagnosed and treated, which includes a galactose free diet. This uncommonly complicates the situation, as galactose is found in most eatables.

Keywords: Galactosemia, GALT gene

The need for interdisciplinary collaboration during the treatment of orthodontic patients

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Introduction: Interdisciplinary collaboration during the treatment of orthodontic patients is an important part of the therapeutic process. During this process the orthodontist should collaborate with other experts in the field, which includes constant interaction and communication with each other as well as setting realistic goals for treatment, which will in the end satisfy the interdisciplinary team as well as the patient. The objective of the research is to examine the frequency of interdisciplinary collaboration of orthodontists and other dental specialists as well as to determine which specialty is mostly collaborated with during the therapeutic process.

Material And methods: The research was conducted in Split. The data, that was collected and analyzed, were 586 orthopantomograms (OPGs) belonging to patients from private orthodontic clinics in Split. The patients were both male and female from 10 to 42 years old. The corpus was collected randomly during March and April of the academic year 2015/2016. After gathering the OPGs, they were analyzed in terms of whether there was potential need for interdisciplinary collaboration with different specialist branches or not. The data were analyzed using the software „MS Excell".

Results: The results showed that out of 586 OPGs, 98 patients (16.72%) needed interdisciplinary collaboration. Out of these 98 OPGs most of them, i.e. 47,96% of cases needed collaboration with oral surgeon, after that comes prosthodontist with 25,51%, then restorative dentist with 14,29%, and last periodontologist with 12,24% of cases. As oral surgery was the most frequently collaborated branch, these cases were analyzed in more depth. The analysis showed that 65,96% of cases included impacted canines, 14,98% impacted second premolars, 4,26% impacted molars, 2,13% impacted lateral incisors, 8,51% of cases included implant placements and 4,26% of cases included cleft palates.

Conclusion: The results and literature overview corroborate the hypothesis that there is frequent need for interdisciplinary collaboration during the treatment of orthodontic patients. The results show that an oral surgeon is the most collaborated specialist during the therapeutic process. Further research with a larger sample and separation of patients by age and gender is necessary in order to obtain a full overview of the problems of interdisciplinary collaboration in orthodontics .

Keywords: interdisciplinary collaboration, orthopantomogram, orthodoncy

Spondyloepiphyseal dysplasia congenita (SEDc) as a form of dwarfism

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Introduction: Spondyloepiphyseal dysplasia congenita (SEDc) is a very rare genetic disorder caused by a mutation in the COL2A1 gene which codes type II collagen. The condition follows an autosomal dominant pattern, although generally occurs as de novo mutation, with no family history of the disorder. As the name of the condition suggests, it results in anomalies of the spine and epiphysis of the long bones. In the clinical picture, the dominating traits are dwarfism, short body trunk, micromelia, platyspondyly. Osteoporosis as well as vision and hearing issues are also possible.

Case report: The case represents a 13-year-old female patient born with the diagnosis of spondyloepiphyseal dysplasia congenita. There are no recorded cases of the disorder in the family. The weight at the time of birth was 3050 g, and the height was 46 cm. The Apgar score was 10/10. Craniofacial deformities were noted after birth. At the age of 1,5 months, the weight was 4350 g, and the height was 50 cm. The status recorded shorter limbs in relation to the torso (length of leg 21 cm, femur 11 cm, lower leg 10 cm), smaller hands and feet, a belly that sticks out, a noticeably bigger head and a short neck. Hip deformity (coxa vara) is present, as well as muscle hypotonia, slower motor development, genua valga and generalized osteoporosis. The densitometry of the spine (L1-L4) finds an average bone density for advanced osteoporosis (Z value -2,5) i.e. 73% of the expected value concerning age. There are no visual and hearing impairments. In 2014 she had a guided growth surgery during which an eight-plate was inserted bilaterally in the zone of the greater trochanter. In 2018 she had an alenthesis extraction surgery. At the last check-up in 2019, her weight was 21 kg and her height 116 cm. The patient suffers from hip, leg and lumbar pain. There is a disproportion between the length of the upper limbs and the torso. The range of motion in all the upper and lower limbs is total. There is hyperelasticity present in all joints. Deviation from the regular kinematics of walking is caused by hip dysplasia, the steps are small, and the walk is slower. In September of 2019, she had a corrective and derotational osteotomy of the left femur which improved the congruence of the femoral head. At the moment the patient is awaiting the same surgery on the right leg.

Conclusion: SEDc is a lifelong disease characterized by degenerative changes. It is cured symptomatically and surgically. Although it cannot be completely cured, a quick diagnosis and intervention can help provide a better life for the people affected by this disease and can reduce the number of orthopedic aids.

Keywords: spondyloepiphyseal dysplasia congenita, dwarfism, genetic

HIV – positive young male patient with bipolar disorder and psychoactive substances addiction

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Introduction: School and adolescent medicine is a branch of preventive medicine that keeps track of not only physical status, but also psychological development of children and young people. A young man shown in this case report is an example of challenges faced by school medicine doctors and shows us that we should discuss more about mental health of young people, as well as problems due to drug and alcohol abuse among them.

Case report: In November 2019, a 22 year-old male patient came to the Department of School Medicine due to systematic check-up. During check-up, young men's physical status was fine, nevertheless, during talk, it has been found out that this young man is living in a non-typical family circumstances, moreover, he has been consuming marijuana since he was 15 years old, as well as ecstasy and amphetamins since he was 18 years old. Patient has been regularly consuming alcohol, often in great amounts. Besides, until now, patient has had around 30 sex-partners (first sexual intercourse was with 16 years), he is homosexually oriented and for the last three years, due to his HIV positivity, he has been taking a medicine which contains three different medications: emtricitabine, rilpivirine and tenofovir alafenamide. In 2017 patient has had his first episode of visual-auditory hallucinations, for which he was hospitalized in Clinical Hospital Centre in Zagreb, when psychiatrist diagnosed him with bipolar disorder and prescribed him psychiatric therapy which he has been taking regularly. Moreover, patient mentioned his grandmother suffered from schizophrenia and his father suffered from PTSD, which led him to commit suicide when patient was 9 years old. After revealing the questionnaire answers about mental health, it is clear that the patient has extremely low self-esteem, along with disrupted image of himself and his values.

After the check-up was performed and the patient was advised about improvement of mental health, but also about benefits of quitting doing drugs, smoking and consuming alcohol, patient was given a new check-up date in two weeks. He was also suggested to turn to psychiatrist or to mental health department, since his symptoms were all connected to mental problems and since depressive episode has occurred.

Conclusion: Despite of activities being done in the domain of school medicine physician, young men's state had not improved. Therefore, these types of patients demand multidisciplinary health care, where psychiatrist and psychologist need to be included, in order to maximize patient's potentials and to bring his own mental health to the highest possible level.

Keywords: alcoholism, drugs, HIV positivity, mental health

Patient with newly diagnosed celiac disease and ataxia – case report

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Introduction: Ataxia is movement disorder characterised with lack of muscle control or coordination of voluntary movements, which could affect various muscles, creating difficulties with stance, gait, hand, leg and eye movement, speech and swallowing. Celiac disease is an autoimmune small intestine disease triggered with gluten, a protein found in wheat, barley and rye. IgA anti-gliadin antibodies are positive in 95 % of patients with untreated celiac disease.

Case report: In 2019, a 49-year-old male patient came to the Department of Neurology due to problems with gait. His symptoms start few years ago when he noticed that he walks on wide base. On sudden sound he experienced involuntary jerks on his hands. Patient's handwriting has changed recently; nevertheless, he did not notice speech problems. He also didn`t notice worsening of the symptoms during the time. His other medical history was negative, same as family history for movement disorders. In neurological examination patient had symptoms of cerebellar ataxia, both on body, hands and legs, with dysarthria and startle myoclonus. Reflexes were symmetrically exaggerated and Babinski sign was noticed on the right foot. Vibration was diminished on the both feet. Muscle strange and light touch were normal, same as cranial nerve innervation. Extensive diagnostic workup was performed (genetic testing on spinocerebellar ataxia type 1,2,3,6,7 and Friedreich ataxia, serology on GAD, vitamin E, paraneoplastic antibodies – Hu, Yo, Ri, serology on autoimmune disease, brain MRI, cerebrospinal fluid) and all findings were normal. Anti-gliadin IgA antibodies were highly positive. We also noticed high level of homocysteine. Psychological testing noticed mild cognitive impairment while electromyoneurography showed sensor axonal polyneuropathy. Patient was referred to gastroenterology and he performed gastroscopy with stomach biopsy. He was advised to start with the gluten-free diet and substitutional therapy of vitamin B12 for a lifetime, as well as folacin through next four months. Patient started with recommended treatment and he is in process of follow up.

Conclusion: It is important in patients with ataxia to think about celiac disease as one of the possible etiology, especially because this condition is possible to treat and to stop further neurological damage.

Keywords: anti-gliadin antibody, ataxia, celiac disease

Congenital mesoblastic nephroma with uncommon evolution

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Introduction: Mesoblastic nephroma is a low-grade fibroblastic neoplasm diagnosed in newborns and infants, generally with a benign behavior. WHO classification mentions a 2 – 4% frequency of all pediatric renal tumors. The prognosis is excellent when the tumor resection is complete, hematogenous metastases and related deaths being rarely reported.

Case study: We present the case of a 4-months old male child, admitted in February 2018 to Pediatric Surgery Clinic of Children's Emergency Clinical Hospital "Saint Mary", Iasi, with the clinical diagnosis: left kidney tumor. After surgical intervention, the excised product was sent to the Pathology Laboratory. The histopathological examination showed a monotone fusiform and ovoid cells proliferation, with high density bundles, interdigitated with renal parenchymal fragments with glomerular immaturity, renal dysplasia with cartilaginous tissue, and subcortical cystic transformation areas. The pathological diagnosis was congenital mesoblastic nephroma. After 3 months, the baby returned to the hospital with a tumor relapse and died after 2 days of hospitalization. The necropsy showed up a left abdominal tumor, infiltrative in the intestine, pancreas and liver. The microscopic examination revealed a cell proliferation with numerous atypical mitosis, a reduced vascular stroma, vascular thrombosis, tumor necrosis and large hemorrhagic areas. The morphological features led to the diagnosis: aggressive congenital mesoblastic nephroma - cell type, with visceral metastases.

Conclusion: This is the case of a 4-month old baby diagnosed with a mesoblastic nephroma with unfavorable evolution. The histopathological exam plays an important role as is the one that sets the diagnosis and guides the therapy, knowing that some variants require more aggressive treatment.

Keywords: Mesoblastic nephroma, Congenital, Relapse, Child

Pulmonary thromboembolism associated with neoplasms

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Introduction: Pulmonary thromboembolism(PTE) is a blockage of an artery in the lungs by a thrombus that migrates from the level of the veins from the lower limbs or it is formed in situ in conditions of hypercoagulability. Patients with neoplasms have a very high risk to develop PTE, but only 10% of the unprovoked PTE are connected to the discovery of a cancer that was initially hidden.

Case study: We present the case of a 65 year-old man who is admitted to the Saint Spiridon Cardiology Clinic with dyspnea at reduced effort and while resting, bilateral limb edema and dry cough which debuted three weeks ago. He is known to have high blood pressure grade 3, chronic hepatitis with virus C and Hodgkin disease with mixed cells. The echocardiography revealed a thrombus in the right atrium and the inferior vena cava, for which thrombolysis is performed. Abdominal ultrasound showed a hyperecogenic nodule, in the right hepatic lobe. On the CT scan appeared the following: mediastinal adenopathies and a hepatoma in the right hepatic lobe, which invades the inferior vena cava and the right atrium through the presence of a quasi-complete thrombosis in the hepatic portion. Hodgkin Lymphoma(HL) is a rare condition and only 20% of the patients with HL have the mixed cells type. The complications of the HL treatment can be solid tumors. Moreover, the chronic hepatitis with virus C is usually linked with Non-Hodgkin's lymphoma, not with HL. 70% of the infections with this virus are becoming chronic, 25% are evolving into liver cirrhosis and only 5% into hepatocellular carcinoma. The main treatment for this patient is low molecular weight heparin anticoagulation for minimum 6 months.

Conclusion: The association of PTE and neoplasms is an unfortunate combination with a high mortality and a high risk of recurrence despite the anticoagulant treatment.

Keywords: Thromboembolism, Tumour, Thrombus, Hodgkin Lymphoma, Hepatopathy

The atrial myxoma and pseudovasculitis syndrome

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Introduction: Cardiac myxoma is a rare benign neoplasm of the endocardium, more common in women as sporadic form or young men as familiar form. From a clinical point of view it is characterized by a triad of cardiac events, embolic events and general symptoms.

Caste Study: We report the case of a 42 year-old male with known history of essential hypertension, type IV hyperlipoproteinemia and previous normal echocardiography. The patient was admitted for gradual onset of myalgia, muscular weakness, prolonged febrile syndrome, 5 kg weight loss and asthenia in the last two months before actual presentation. The clinical exam was normal excepting the persistent fever and pale teguments and mucosa, while the biochemical and hematological parameters revealed a systemic inflammatory response and mild anemia. We conducted the investigations in order to confirm the hypothesis of polymyositis or polyarteritis nodosa. Searching for asymptomatic organ damage in hypertension was also performed. The nerve conduction velocity provided an equivocal pattern for polymyositis while the musculo-cutaneous biopsy was highly suggestive for diagnosis. At the same time, other causes of secondary myositic syndrome were excluded by specific techniques. Renal arteriography revealed multiple microaneurysms suggesting polyarteritis nodosa. Corticotheraphy was initiated with favorable clinical course but persistent systemic inflammatory response. The conclusive exam was transthoracic echocardiography that discovered a left atrial myxoma. The patient was urgently addressed to cardiac surgery for its removal. The short and long-term evolution was uncomplicated.

Conclusions: This case is particular because of the diagnosis circumstances, on the basis of so-named pseudovasculitis syndrome in absence of cardiac manifestations and previous normal echocardiography in a male patient. At the same time, the case highlights the importance of echocardiography during the work-up for a prolonged febrile syndrome.

Keywords: pseudovasculitis, myxoma, febrile syndrome

Pheochromocytoma: a case report

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Introduction: Pheochromocytoma is a tumor arising from chromaffin cells of the adrenal gland medulla and sympathetic ganglia that releases catecholamines.

Case report: The clinical photograph shows a 62-year-old patient having arterial hypertension, type 2 diabetes, and hypercholesterolemia, who had a reduced appetite for three months, lost 10 kg, suffered from pain in the lumbosacral spine and felt depressed. The abdominal ultrasound showed an inhomogeneous tumor mass on the right adrenal gland, 5cm x 4.5 cm in size, with minor cystic spaces in the middle. The computed tomography (CT) scan of the abdomen showed a sharply defined heterogenous mass on the right adrenal gland of 4.2x5 cm in diameter with higher absorption coefficients on the edge and lower in the middle (central necrosis), measuring a density of 26 Hounsfield units (HU), on native non-homogeneous post-contrast opacification sections, showing a slow flush of the contrast on post-contrast sections (a relative wash out amounting to 9%, and the absolute one accounting for 20%) a pheochromocytoma is clinically suspected. Medical records indicate that during the previous four years the patient occasionally suffered from palpitation and supraventricular tachycardia with blood pressure levels up to 190/100mmHg. The laboratory analysis showed elevated plasma levels of metanephrine and normetanephrine (12 and 9 times), confirming the diagnosis. A pre-opeartive preparation was performed using alpha-adrenergic receptor blockers (phenoxybenzamine), followed by beta blockers (bisoprolol). Following the surgery, the histopathological examination confirmed the pheochromocytoma diagnosis. The metanephrine and normetanephrine follow-up results were normal. Annual follow-up checkup is recommended for the next 10 years.

Conclusion: In case of a clinically suspected pheochromocytoma or during the adrenal gland incidentaloma CT imaging the adenoma is excluded (including a density of more than 10 HU), a biochemical analysis of pheochromocytoma must be performed.

Keywords: adrenal tumor, pheochromocytoma, arterial hypertension, palpitation, tachycardia

Multimodality imaging in cardiomyopathies: friend or foe?

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Introduction: Dilated cardiomyopathy (DCM) involves the presence of an enlarged left ventricle/heart chambers with subsequent systolic dysfunction in the absence of coronary artery/valvular heart disease or hypertension. Cardiac multimodality imaging is being increasingly used for identifying the etiology, while a selected number of patients undergo cardiac resynchronization therapy (CRT). We present you a case of an idiopathic DCM male patient who received CRT after remaining symptomatic despite adequate medical therapy.

Case study: A 40-year old male patient presented with aggravating dyspnea and frequent palpitations for the previous 6 months. He was diagnosed with a left bundle branch block 4 years prior to current admission and with a myocardial infarction 3 months earlier, when transthoracic echocardiography revealed a left ventricular ejection fraction (LVEF) of 20% and an end-diastolic left ventricular diameter of 70 mm. The aetiology of DCM was considered to be ischemic at that time. Coronary arteries catheterization revealed normal coronary arteries. Cardiac magnetic resonance (CMR) raised the suspicion of cardiac sarcoidosis, the second possible cause of the DCM. However, the latter was infirmed when the patient was implanted with a CRT-D (implantable cardiac resynchronization therapy defibrillator).Evolution was favourable, the patient being a super-responder with a 20% increase in LVEF 3 weeks post-implant.

Conclusion: CMR is used for its tissue characterization abilities, namely in identifying fibrosis' presence and pattern. However, these patterns are interchangeable and there is a lack of consensus in diagnostic criteria. As such, interpretations should be considered as highly suggestive and not clear diagnostic and be judged in the clinical context. Treating DCM is a challenge and multimodality imaging, in the absence of a holistic approach may rather become a foe instead of a friend.

Keywords: cardiomyopathies**,** systolic dysfunction

Facial dysmorphism and intellectual disability in a rare 19q13.43 microdeletion syndrome

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Introduction: 19q13.43 microdeletion syndrome is a rare genetic disorder arising from a non-recurrent rearrangement including peculiar facial features, intellectual disabilities, delayed growth and development, and feeding difficulties. Most patients have impaired EEG and epilepsy.

Case report: This case represents two children with 19q13.43 microdeletion syndrome. First, female child of healthy nonconsanguineous parents, born in the second pregnancy with birth weight (BW) 2850 g, Apgar score (AS) 10 and postnatal presenting with hypertonia. Early psychomotor development was delayed and moderate intellectual disability (IQ 45). Epilepsy occurs at the age of 16 months. Dysmorphic features include the long face, epichantal folds, synophrys, gothic palate, irregular growth of teeth, maxillary prognathism, low set hair on the neck and head, hypermobile joints, long, thin fingers, clinodactyly. Magnetic resonance imaging (MRI) showed ventriculomegaly and hypoplastic corpus callosum. A karyotype is normal with heteromorphism of chromosomes 16. Multiplex ligation-dependent probe amplification (MLPA) found microdeletion 19q13.43. The girl now has 20 years, without epilepsy, delayed cognitive development and attention deficit. Second, 15 years old boy of nonconsanguineous parents, born in the second pregnancy BW 3,4 kg, AS 10. Positive psychiatric heredity and exposition to family violence. As in the first case, epilepsy and phenotypic feature are present, differences are hypertelorism, large ears with otapostasis, convergent strabismus, pectus infundibuliform, umbilical hernia, and scoliosis. He has border cognitive functionating, hyperactivity, attention deficit disorder with autistic behavior. MRI showed expand liquor space at the base of the brain. A karyotype is normal. With the MLPA method microdeletion 19q13.43 was found.

Conclusion: This rare microduplication syndrome includes several genes such as SCN1B, whose haploinsufficiency is associated with epilepsy and febrile seizures. Other deleted genes are from KRAB-ZNF clusters which act as transcription factors in brain differentiation and in development of cognitive functions. There is no cure for 19q13.43 deletion syndrome it is treated symptomatically. Affected individuals need a team of specialized doctors for treating the various problems.

Keywords: Multiplex ligation-dependent probe amplification (MLPA), microdeletion, 19q13.43, intellectual disability, epilepsy

Risk Factors for Laryngeal Cancer

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Introduction: Laryngeal cancer is one of the most common types of head and neck cancers and represents 1-2% of all malignancies. The vast majority (95%) of cancers of the larynx are squamous cell carcinomas that arise from the covering of the vocal cords. Tobacco is known to be a dominant predisposing factor and smokers carry 10-20 times greater risk for developing this type of cancer. It is known that giving up smoking rapidly decreases the chances of getting cancer. This is why it is important to make sure that diagnosed patients stop smoking. Alcohol is another predisposing factor, especially when combined with tobacco. Laryngopharyngeal reflux and human papillomavirus (HPV) are mentioned as possible risk factors as well. The most common cancer localization, where HPV antigens were found, were the tonsils (74%), larynx (30%) and tongue (22%). Almost every head and neck tumor that is HPV positive is HPV type 16, which usually incorporates into the genome of the host. In addition to that, there is another etiological factor known to be the cause of HPV infection and laryngeal cancer, and that is unresponsible sexual behavior.

Case report: History. A 34-year old male patent with a hoarse voice that changed in intensity and has lasted for a month, before he was admitted to the hospital. The patient denied difficulties with swallowing and breathing. In the last 15 years, he has been constantly exposed to voice effort (he had spent 8 hours daily training the kids in the football academy). He is a nondrinker, a nonsmoker, denies drug usage and other comorbidities. In 1996 he underwent an appendectomy. He is not married and lives with his fiancé. When admitted, he was afebrile, with bodyweight – 83kg, body height – 190cm, and RR- 130/80 mmHg. Rhinoscopy showed that mucus in the nose cavity was pink, thickened, and showed deviation on the left. Oropharyngeal examination showed red mucous on the pharynx with uneven tonsils, reaching above the palatine arches. In the front 2/3 of the left vocal cord, there was a papillomatous exophytic growth, 1 cm in size, and on the right vocal cord, there was a smaller cystic formation. Both vocal cords were movable during phonation and respiration but incompletely adducted because of the growth on the left vocal cord. Bilateral otoscopy showed a settled triangular reflex. The neck was palpably free. Microlaryngoscopy and biopsy were done, and histopathological results confirmed HPV positive squamous cell carcinoma. Expanded cordotomy was done, and since then, the patient undergoes LMSC and neck ultrasound regular checkups, visits speech therapist and a psychiatrist (he developed anxiety and fears the progression of his disease).

Conclusion: In the last couple of years, unresponsible sexual behavior and HPV infection became a major worrying cause of head and neck cancer. The number of young people suffering from laryngeal cancer is increasing, where the main etiological factor becomes oral intercourse. The best way to avoid this type of cancer is to educate young people about HPV infection before they become sexually active and to show them healthy lifestyle options.

Keywords: HPV, laryngeal cancer

Simultaneous floating thrombus in the ascending aorta and pulmonary embolism

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Introduction: Thrombi originating from the ascending aorta occurring simultaneously with bilateral pulmonary embolism are rare. The association of arterial and venous thrombosis is also very uncommon.

Case study: We present a case of a 83-year-old woman with a history of diabetes mellitus, hospitalized for aggravating dyspnea associated with congestive heart failure, chronic kidney disease, paroxysmal atrial fibrillation, and severe hypoglycemia. The latter was corrected in the emergency room. A floating thrombus of 22/37 mm in the ascending non-dilated aorta (of 24 mm) was incidentally discovered during a computer tomography (CT) angiography for a high clinical suspicion of pulmonary embolism. Thoracic CT scan also showed bilateral pulmonary embolism in inferior pulmonary lobes. Logistic Euroscore was 51.70 %. After 24 hours of conservative treatment, the multidisciplinary heart team decided to continue with non-fractioned heparin in intensive care unit. After 2 weeks of congestive heart failure treatment optimization and heparin therapy, the repeated CT scan revealed an irregular aortic intimal surface, while the floating thrombus in the ascending aorta was no longer detected. The presence of a thrombophilia has yet to be confirmed by blood tests and neoplasia was not diagnosed.

Conclusion: Currently, the guidelines offer no consensus on how to treat a free floating thrombus in ascending aorta. Therefore, such cases represent a challenge in what concerns management and appropriate treatment.The severe dehydration associated to procoagulant status of chronic kidney disease and paroxysmal atrial fibrillation probably determined simultaneous arterial and venous thrombosis.

Keywords: floating thrombus, pulmonary embolism

Takotsubo cardiomyopathy presenting with ST elevation in patient with an end stage renal disease on hemodialysis program: case report

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Introduction: Takotsubo cardiomiopathy (TC) is a transient cardiac syndrome that involves left ventricular apical akinesis and mimics acute coronary syndrome. Chest pain is the most common presenting complaint among patients, second to dyspnoea, and other less frequent symptoms include syncope, cardiogenic shock and cardiac arrest. It occurs more often in postmenopausal eldery women, associated with emotional or physical stress and its presented with chest pain, ST-segment elevation on electrocardiography and elevated levels of cardiac enzymes. Diagnostic criteria were published by Mayo Clinic in 2004. and reevaluated in 2008., with the addition of new criteria.

Case report: We report the case of 83 year-old woman with an end stage renal disease on hemodialysis program (HD), who was hospitalized due to a gram-negative sepsis, which was confirmed with blood culture and laboraty findings. On the 7th day of hospital treatment, the patient complained of chest pain and shortness of breath. The chest pain traveled up to the neck, into the jaw, and then radiated to the back, with no vomiting or swelling. An electrocardiogram showed sinus tachycardia with left electric axis. An ST elevation was observed of 1,5 mm in anteroseptal leads, not previously described. Laboratory findings showed a significant increase in cardioselective enzymes – troponin hsI 523,2 ng/L (upper limit of normal less than 15,7 ng/L). Departmend of cardiology was consulted, and the patient was transferred to Coronary care unit (CCU). Cardiac catheterisation laboratory (Cath lab) was activated and patient underwent coronary angiography which showed normal findings and no signs of obstructive coronary artery disease. Furthermore, transthoracic echocardiography was performed which verified left ventricular akinesis, predominantly apical (LVID 4,6 CM, IVSd 0,8 cm) with large apical thrombus. The ejection fraction by Teichholz formula was 35%, mitral and aortic valves were normal, with no pericardial effusion and no pulmonary hypertension. Follow-up electrocariogram showed atrial fibrilation, wirh resolution of the ST elevation. Patient was treated with warfarin anticoagulant therapy with prothrobin time control and other symptomatic therapy. after 20 days of hospital treatment, the patient was discharged and continued with HD treatment.

Conclusion: In conclusion, we present a case of Takotsubo cardiomyopathy in patient with an end stage renal disease on HD program. According to available data, there were only 30 cases of TC described which shows that TC is a rare cardiomyopathy in an end stage renal disease.

TC should be considered in the differential diagnosis in hemodialysis patients, particularly who present with chest pain and/or symptoms.

Keywords: cardiomyopathy, renal disease

The importance of accurate diagnosis in case of stress and urge incontinence

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Introduction: Urinary incontinence is a condition characterized by uncontrolled leakage of urine. It usually occurs in the elder population, but it is women who are most affected by this abnormality. There are two most common types of urinary incontinence – stress (static, SSI) and urge incontinence. The difference between these two types lies in their cause. Stress incontinence is caused by physical activity that increases bladder pressure, such as sneezing, coughing and heavy lifting, while urge incontinence is the result of overactive bladder. The treatment is similar for both of these types (bladder training, pelvic muscle exercises), but it is important to emphasize that surgical approach is only an option in stress incontinence.

Case report: We present the case of a 53-year-old female patient suffering from stress incontinence. She was admitted to the hospital, claiming that she noticed involuntary urine drainage in the last 3 years, which leaks even during less physical exertion. Gynecological examination was performed to evaluate external genitalia as well as the uterus, fallopian tubes and ovaries, which seemed neat. After the Bonney test, which turned out to be positive, the patient was diagnosed with stress incontinence. Due to the fact that patient was diagnosed with SSI, she underwent surgery, in which was used periurethral gel application based on a hyaluronic acid.

Conclusion: Since we differentiate two types of urinary incontinence that share similar symptoms, it is important to establish an accurate diagnosis in order to ensure an appropriate treatment.

Keywords: urge, stress, incontinence

The role of heredity in Bipolar affective disorder

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Introduction: Bipolar disorder is a common disorder characterized by unpredictable changes between mania and depression. Considering the fact that the treatment, if diagnosed early, can be very successful, numerous attempts have been made in order to discover genetic risk factors or biomarkers that could help identify individuals at risk. Genetic factors (predisposition) and environmental risk factors such as alcohol or drug dependence, trauma or even metabolic diseases all in combination are triggers for the beginning of the disorder. Considering all genetic risk factors the most important are definitely single nucleotide polymorphisms (SNPs) in the genes CACNA1C, ODZ4 and NCAN.

Clinical Case: A 62-year-old patient was admitted to the Department of Psychiatry due to the worsening of her symptoms. She is in a psychiatry treatment since 1993. under the diagnosis of Bipolar affective disorder. The patient is widowed, mother of two, retired and currently living with her son. On the day of admission, the patient presented with psychomotor slowing, impaired affect modulation, and a thought dissociation. A week after the patient was admitted we conducted an interview with her. The first noticeable thing was her drawings all over her skin and pyjama. The patient admits hearing voices in the form of a murmur and having illusions. During our interview, the patient's thoughts were dissociated and disrupted by delusions. Like many other patients diagnosed with bipolar disorder, she tried to commit suicide, according to her, on two occasions. Apart from Bipolar affective disorder, she also suffers from hypothyroidism. The last thyroid function blood test was made in July 2019. and TSH, fT3, and fT4 were all within reference values. During the last hospitalization, the patient was treated with aripiprazole, olanzapine, lamotrigine, clonazepam, biperiden, flurazepam, phenothiazine and promazine. Hypothyroidism was treated as well with levothyroxine sodium.

Discussion: With this case study, we wanted to show some of the most important genetic risk factors as well as some metabolic disorders involved in the development of the bipolar affective disorder. Our patient suffers from hypothyroidism. Some authors suggested that rapid administration of thyroxine could lead to the catecholamine receptor sensitivity leading to hypercatecholaminergic state. Our patient was treated with low dosage of levothyroxine, but her thyroid functional parameters were within reference values showing that the treatment of hypothyroidism was not the trigger for mania.

Keywords: Bipolar disorder, hypothyroidism, genetic risk

The unspoken need of blood products required in acute aortic dissection

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Introduction: Acute aortic dissection is a major emergency, involving extracorporeal circulation and increased duration of intervention which makes transfusion therapy essential in the management of the patient. The aim of this study is the identification of particular aspects of transfusion therapy in patients with aortic dissection who have undergone surgery, as well as the analysis of blood requirements, choice of blood products and compatibility in patients.

Materials and methods: This retrospective study included the analysis of the observation sheets of 50 patients who had surgery for acute aortic dissection in the Institute of Cardiovascular Diseases Iasi (Jan 2016-Dec 2017).

Results: The average age of the patients was 58 years, with 62% men and 38% women. The maximum operating time was 20 hours (average 9.52 hours). The duration of mechanical ventilation was 171.44 hours, and most patients (62%) were hospitalized between 11 and 30 days. The consumption of blood products had an average of 3610 ml (20.5 units), with a maximum of 11260 ml (74 units) as follows: packed red blood cells - average of 2144 ml (max. 6500 ml); fresh frozen plasma - average 1391 ml (max. 4950 ml); platelets - average 160 ml (max. 650 ml); cryoprecipitate of factor VIII: average of 65 ml (max. 460 ml). The study showed that 27% patients needed a volume > 5000 ml of blood products (the maximum consumption was 11260 ml with an average of 3610 ml). The most frequent group was A, followed by groups O, B and AB. For patients with Rh negative (10.41%), blood supply was difficult. 27% of patients received massive transfusions.

Conclusion: The mortality of the patients with acute aortic dissection admitted to our institute was 23%, indicating that the chances of survival increase with immediate intervention and availability of high amounts of transfusion products.

Keywords: aortic dissection, blood, mortality

Transient ischemic attack - the first manifestation of a rare genetic disorder

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Introduction: Hereditary Hemorrhagic Telangiectasia (HHT) is estimated at 2.0 per 10,000 persons worldwide,being characterized by the presence of muco-cutaneous telangiectasia and visceral arterio-venous malformations.

Case study: A 39 years old male with a history of recurrent anterior epistaxis, telangiectasias on lips and nose from the age of 7 is presented for epistaxis, dyspnea, signs and symptoms for transient ischemic attack. Chest x-ray and blood oxygen levels are not sensitive enough to detect the PAVMs in this way we need to use chest computer tomography scan, pulmonary angiography or contrast echocardiography.Clinical examination and the transthoracic echocardiography were normal. Arterial blood gas exam detected an oxygen saturation of 98% which decreased at 89% in walking condition. Contrast transthoracic echocardiography showed a massive right to left late shunt compatible with the presence of intrapulmonary shunt quantified at 15%. The CT and angiography confirmed the presence of a pulmonary arterio-venous malformation (PAVM) on the inferior right lobe. The cerebral MRI detected the presence of old and recent ischemical lesions on the left cerebellum hemisphere. The radiological and endoscopic evaluation did not detect other AVM on the brain or gastrointestinal tract.Similar PAVMs were detected to other 5 family member from 7 investigated, 3 of them presenting less severe respiratory, nose and throat symptoms. Genetic test showed a genetic disease with autosomal dominant transmission and incomplete penetration,the Rendu-Osler-Weber disease(known as Hereditary Hemorrhagic Telangiectasia).

Conclusion: Hereditary Hemorrhagic Telangiectasia is a rare condition, usually underdiagnosed and associated with multiple treatments challenging for the specialists. Even is an autosomal-dominantly inherited disease, the manifestation are not present from birth but develop with increasing age.Also, pulmonary AVMs provide a direct communication between pulmonary and systemic circulations, and are often multiple, bilateral and located at the bases of the pulmonary lobes. The PAVMs have a tendency to increase in size over time and puberty, pregnancy and pulmonary arterial hypertension can explain this phenomenon. Routine screening for PAVM's for the patients diagnosed with this condition is indicated and can prevent severe complications as brain abscess and stroke.

Keywords: ischemic attack, genetic disorder

Translocation Patau syndrome with long survival and comorbidity

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Introduction: Chromosomal aberations can have a recognizable pattern of congenital development anomalies followed by a high mortality rate early in life. Patau syndrome is caused by trisomy of chromosome 13, which ocurrs most commonly in mothers older than 35. Secondly, Robertsonian translocation which produces an additional long chromosome arm. The rarest type is mosaicism, where some cells have trisomy. A typical clinical image is a set of respiratory, cardiovascular and digestive system abnormalities. Most children die within the first week of life. Survival after the first year is rare, which depends on the complexity and the possibility of treatment.

Case report: We are presenting a case of a 12-year old girl with Patau syndrome, monitored from birth to her current age. She is the first of three children of young and healthy parents. The patient has most of the phenotypic traits characteristic for the Patau syndrome. Microcephaly and anopthalmia are present. An MRI showed typical malformations: cortical atrophy and hypoplasia of the corpus callosum and the cerebellar vermis. The anatomical structure of the auricle is irregular; the patient also has arachnodactyly and a foot anomaly: the calcaneus is prominent distally with a convexity across the lenght of the foot. A severe cardiac defect is shown as well; ventricular septal defect is present and the aorta originates from the right ventricle due to the absence of the interventricular septum. Throat musculature atrophy is present and because of that the patient is not able to swallow and has no fonation ability. The child requires nasogastric tube feeding and is entirely dependent on the parental care.

The correction of the heart malformation was successfully performed in 2008. A control ultrasound scan is clean. The patient is frequently hospitalized due to recurrent respiratory and urinary infections. Epilepsy has developed in the age of 8 and it is exceptionally treatment – refractory. The satisfying therapeutic effect is noticed only when 5 different antiepileptic drugs are applied. The patient is currently being prepared for the spine deformity operation (scoliosis).

Conclusion: Patients can have longer life expectancies in cases of milder clinical expression of symptoms, like in the translocation typ. Every patient is particular and when deciding about treatment, all comorbidities should be assessed and parent's choices respected. Treatment should be directed towards both treating the patient and supporting parents, as well as genetic consultations when planning future pregnancies.

Keywords: translocation, Patau, genetic

Triple X syndrome as a cause of cognitive impairment

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Introduction: Triple X syndrome (trisomy X or 47,XXX) is a rare genetic disorder that affects females and occurs due excess of X chromosome. Majority of females don't experience any of the symptoms, but in others they can be apparent and including developmental delays, learning and intellectual disabilities. Also seizures and kidney abnormalities can occur. Other most common features involve rapid growth and behavioral problems such as ADHD. Females with triple X syndrome are phenotypically indistinguishable among other females. Treatment for triple X depends on it's symptoms and severity.

Case report: We present a case of triple X syndrome in two females. First, female child of healthy nonconsanguineous parents born in a second pregnancy. Birth was in term, naturally with prolonged leakage of fertile water and revivification (they spent 12 days in hospital). At the age of 7 she was sent at endocrinological outpatient clinic for the appearance of pubic hair and rapid growth. Her mother also noticed a lack in impaired logical reasoning. Karyotyping confirmed triple X syndrome (karyotype: 47 XXX). The girl now has 8.5 years and is showing some features of the syndrome: rapid growth, round head, wider ears, mild hypertelorism, breast Tanner 1, without glandular tissue, axillary hairs Tanner 1, pubic hairs Tanner 2. The second case represents a 12-year-old girl who also suffers from rapid growth and a milder impaired cognitive functioning. Karyotyping confirmed triple X syndrome. Currently she takes Lamotrigine due to EEG changes. She never had epileptic seizures. Both patients were sent to make gynecological examination, UZV and were sent to psychologist for assessment of cognitive ability.

Conclusion: Triple X syndrome characterized by low intellectual abilities and delayed development of motor and speech skills is hard to recognize because of it's mild symptoms and hardly existent physical differences. Therefore it is important to make a diagnosis and set a treatment depending on symptoms needs, but syndrome itself has no cure.

Keywords: Triple X syndrome, cognitive

Unusual retinal hemorrhages as a first sign of chronic myeloid leukemia: a case report

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Introduction: Chronic myeloid leukemia (CML) is a myeloproliferative clonal hematopoietic disorder characterized by a specific reciprocal translocation of c-abl gene from chromosome 9 to chromosome 22. C-abl attaches itself to bcr gene and creates fusion gene bcr-abl responsible for overexpression of tyrosine kinase. Twenty to 50 percent of patients are asymptomatic, with the disease first being suspected from routine blood tests. Among symptomatic patients, symptoms of anemia, splenomegaly, and hyperleukocytosis are common. Initial presentation with visual disturbance, without other symptoms, is very rare in CML. We report a case of CML presenting with retinal hemorrhages.

Case report: A 34-year-old man, without any comorbidities, presented with complaints of blurring in right eye. He did not have a history of other symptoms of neurological disturbances or bleeding manifestations. The visual acuity was 0.5 in the right eye, with normal vision on the left. The anterior chamber was normal. There was intraretinal hemorrhage in the right eye, confirmed by fundoscopy and optical coherence tomography (OCT). A hemogram showed a total leukocyte count of 178,3 x109/L, hemoglobin of 112 g/L, and a platelet count of 238 X109/L. Differential leukocyte count was suggestive of left shift with prominence of myeloid precursors: polymorphs 54%, lymphocytes 6%, eosinophils 4%, basophils 4%, metamyelocytes 11%, myelocytes 12%, promyelocyte 3%, and myeloblasts 4%. He had a splenomegaly of 5 cm below left costal margin. There was no lymphadenopathy or bleeding manifestations. The bone marrow was suggestive of CML chronic phase. Conventional cytogenetics revealed a karyotype of 46, XY, t (9;22). FISH analysis proved Philadelphia chromosome in 96% of cells. Qualitative reverse transcriptase polymerase chain reaction (RT-PCR) for bcr-abl1 translocation showed fusion transcript of b2a2 encoding for major transcript p210. He was classified as Sokal, Hasford intermediate risk, and EUTOS low risk and was started on imatinib 400 mg orally once daily. One month post imatinib, his visual acuity improved to 1.2 in right eye and fundus examination and OCT showed resolving fundal hemorrhages. At 6 months, bcr-abl quantitative analysis showed 0.825275 %. He attained complete cytogenetic remission after 6 month of imatinib.

Conclusion: CML should be considered as differential diagnosis in patients with retinal hemorrhages. Visual disturbance, even they are rare in CML patients, should not be neglected as they can be managed by TKIs without the need for local therapies.

Keywords: Chronic myeloid leukemia, retinal hemorrhages, hematopoietic disorder

Very long-chain acyl-CoA dehydrogenase deficiency – case report

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Introduction:Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency is a condition that prevents the body from converting certain fats to energy, particulary during periods without food. Signs and symptoms of VLCAD deficiency typically appear during infancy or early childhood and they are manifested by hypoketotic hypoglycemia with possible seizures, vomiting, weakness, impaired consciousness, respiratory failure, liver abnormalities, life-threatening heart problems and distruction on muscle tissue.

Case report: A 6-year-old male patient, born at 41st week from two young, healthy and nonconsanguineous parents, began to present specific clinical manifestations of this enzyme disorder immediately after birth. The first minute after induced labor, Apgar was 5/10, the child generalized livid, with hypotonia, tachycardia, infrequent respiration finished by oxygen during 4 days in an incubator of intensive care department. Initial acid-base status and laboratory findings : mild respiratory and metabolic acidosis, lactate = 4.8 mmol/L, AST = 411 U/L, ALT = 83 U/L, GGT = 97 U/L, CK = 14790 U/L, LDH = 2028 U/L. Ultrasound revealed diffuse hyperechogenic liver and echocardiography showed ASD II. A disorder of long-chain fatty acid breakdown was suspected when urine analysis showed increased excretion of dicarboxylic acids. Condition was confirmed based on DNA extraction from a blood sample and findings of undetectable acyl-CoA dehydrogenase very long chain enzyme activity in fibroblast culture.

Due to proper and regular diet, which implies reduced intake of long-chain fatty acids and sufficient use of essential fatty acids, there were no recurrent episodes of muscle weakness, liver enzymes have normalized and repeated echocardiography revealed concentric non-obstructive cardiomyopathy with patent foramen ovale, along with no life-threatening arrhythmias detected on the holter ECG.

In the first three years of life, with more frequent respiratory infections, he had been developing metabolic crises, which were treated in hospital. Now, at the age of seven, starting an elementary school has been postponed due to autism spectrum disorder and is currently in process of rehabilitation.

Conclusion: Inherited metabolic disorders are group of rare disease. Treatment is available if they are recognized promptly. VLCAD can be treated with an adequate nutrition and repression of metabolic crises. Since 2017, in Republic of Croatia, VLCAD has been included in a newborn screening test which led to detecting four different cases.

Keywords: inherited metabolic disorders, hypoglycemia , very long-chain acyl-CoA dehydrogenase deficiency

Perioperative managment of patients receiving direct oral anticoagulants in dentoalveolar surgery

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Introduction: The number of geriatric patients seeking dental practice is ever-rising because of increased life expectancy, also with problem of increased medical conditions. One of very common patients in dental clinics are patients on novel oral anticoagulants (NOACs). These patients present a challenge due to their increased risk of bleeding. Hemostasis is crucial for success of oral surgical treatment so aim of the paper is to optimize the management of the dental patients on novel oral anticoagulants in a standardization of treatment protocol.

Review: New oral anticoagulants are also aclled „non-vitamin K antagonist oral anticoagulants“ and „directly acting oral anticoagulant“. They exhibit rapid onset time and prompt withdrawal of anticoagulation effect when set because of their rapid action and short half-lives. These more flexible pharmacokinetic properties allow short-term interruption without increasing the risk of relapsing thrombotic or cardiovascular events. Before providing dental treatment for a patient taking novel oral anticoagulants their bleeding risk must be assessed. This paper involves consideration of both likely risk of bleeding associated with the required dental procedure and patient`s individual level of bleeding risk presented in the tables. Dental procedures that are likely to cause bleeding are categorized in table as either low-risk or high-risk procedures. Tables in the paper intend to be a guide and bleeding risk should be judged based on individual case. Careful patient selection helps preventing bleeding complications.

Conclusion: The perioperative management of patients using novel oral anticoagulants who undergo dental procedures represent a common clinical problem. Therapy with new oral anticoagulants brings new challenges for dental practioners assessing the risk versus benefit of cessation versus non-cessation of anticoagulant therapy for dental treatment. Further studies are needed to establish evidence-based guidelines fot the perioperative treatment of these patients.

Keywords: anticoagulants, dentoalveolar surgery

Cancer organoids as preclinical model for drug development

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Introduction: Cancer mortality rates remain high worldwide, largely due to inefficient treatment often caused by resistance to existing therapies. Hence, new anticancer therapies are urgently needed, but the development of such therapies is slow and expensive, mainly due to the shortcomings of currently used preclinical screening models. Therefore, it is crucial to find the most appropriate model by which effective chemotherapies could be found. Cancer organoids, which are the focus of this review, are seen as a promising preclinical model for drug discovery.

Review: When testing new anticancer drugs, in vitro cell cultivation is an inevitable technique. Traditionally, two-dimensional (2D) cell cultures have been the most commonly used model, but they lack in vivo features such as the extracellular matrix and cell-cell interactions. Hence, the results obtained from 2D models are often inaccurate, showing hypersensitivity to clinically ineffective oncology drugs. Patient-derived xenografts (PDX), obtained by implantation of a patient's tumor tissue in immunodeficient mice, are a widely used model in pharmaceutical research. PDXs closely mimic tumor histology and therapy response but are costly, time-consuming and not suitable for high-throughput screening. To mirror the microenvironment of the native tumor tissue and to model drug responses, various three-dimensional (3D) in vitro models have been developed. Cancer organoids, miniaturized 3D self-organizing tissue models that mimic the parent tissue or organ, are a promising model for pharmaceutical research, especially in personalized medicine when cells are derived from patients. In this way, they could be used to predict the patient's response to medication. Organoids are easy to initiate, are suitable for high-throughput assays and have the characteristic of tumor heterogeneity. Although they have many benefits compared to 2D and PDX models, organoids lack the vascular and immune system, as well as stroma which makes them insufficient to be a sole model in drug research.

Conclusion: Cancer organoids hold great potential as a reliable drug discovery system. They closely mimic tumor organization in vivo, provide fast and high-throughput results, and thus fill the gap between simple 2D and complex PDX models. However, future developments such as recreation of vascular system and incorporation of immune cells are needed to accurately represent the in vivo drug response. Overall, the advancement of organoid cultures could provide us with a predictive model that would make drug development much more efficient and allow a personalized therapy choice.

Keywords: cancer organoids, drug model, tumor

Review of recent advances in preimplantation genetic diagnosis

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Introduction: Preimplantation genetic diagnosis (PGD) was introduced in the late 1980s to fertile couples at risk of transmitting X chromosome-linked diseases. During the last decade, PGD has evolved to indicate a wide range of inherited genetic diseases, mutations in the DNA, and aneuploidies especially in women of advanced maternal age (AMA).

Review: Today it is more often used as a screening method for euploid, healthy and viable embryos during the in vitro fertilization (IVF). As synonyms are used preimplantation genetic screening (PGS) and preimplantation genetic testing (PGT), however, PGT is referred to testing for aneuploidies.

Whiles, there are several proposed methods, it is still debating between non-invasive and invasive testing. Invasive procedures, i.e., trophectoderm, blastomere and polar body biopsy require removal of the cells from preimplantation embryos and it's considered that they have an adverse impact on the development of an embryo. However, it has been proven that the use of laser manipulation on biopsy karyotype doesn't have a negligible impact on PGS results, but it is yet unknown how it can affect the embryo viability. Meanwhile, several new procedures were introduced into the field including time-lapse technology, blastocoel fluid sampling and cell-free nucleic acid collection from the spent culture medium. These methods have limitations and require further examination and validation for their clinical use. Furthermore, it has been proven that results about aneuploidy obtained from the spent culture medium and blastocoel fluid differ. Thankfully to the rapid development of PGD/PGS, there are reports of a few cases of healthy baby deliveries. For example, while the mother is a carrier of X-linked disease, mucopolysaccharidosis type II, the male baby was born healthy. In addition to that, PGT is a successful method to determinate aneuploid pregnancy of women of AMA that undergo IVF. According to research conducted on 627 women, clinical pregnancy rates were higher after elective single embryo transfer versus non-elective.

Conclusion: We conclude that PGD/PGS is an appropriate reproductive option for couples at risk of transmitting monogenic diseases, as well as PGT is for women of AMA at risk of aneuploidy pregnancy during IVF. PGT can be safely offered to all patients to improve implantation rates, reducing miscarriage and multiple pregnancies. Studies of the viability of all embryos that undergo the PGD/PGS/PGT need to be expanded due to the possible negative effect of procedures.

Keywords: preimplantation genetic diagnosis, in vitro fertilization

A Review on Platelet Activating Factor Inhibitor Lexipafant: Could Lexipafant be a Standard in the Treatment of Sepsis?

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Introduction: Sepsis is a serious public health problem despite the use of modern antibiotics and resuscitation therapies. Every year, sepsis affects 20-30 million people and 7 to 9 million die – which means one death in every 3.5 seconds. It is still the most common cause of death among critically ill patients. There are numerous clinical trials for a new non-antibiotic treatment for sepsis and one of them is Lexipafant.

Sepsis and Lexipafant: According to The Third International Consensus Definitions for Sepsis and Septic Shock it is defined as a life-threatening organ dysfunction caused by a dysregulated host response to infection. Organ dysfunction can be identified as an acute change in total SOFA score. The most common cause are gram-negative organisms. Clinical features of sepsis are very variable. Platelet-activating factor receptor is a member of G-protein coupled receptor superfamily. Recent studies have shown that immune system, during the septic response, produces proinflammatory cytokines and PAF in large amounts. Lexipafant is a potentional PAF antagonist and previous research showed positive effects in acute pancreatitis. Acting and treating acute pancreatitis reduces the risk of developing sepsis. Yupin Suputtamongkol et al. investigated effect of Lexipafant in 131 patiens with sepsis, but in the end study showed that Lexipafant was well tolerated but not associated with a significant reduction in mortality.Other study conducted on a patients with severe acute pancreatitis that led to organ failure and sepsis, showed that Lexipafant had no effect on new organ failure during treatment what would lead to conclusion that antagonism of PAF activity on its own is not sufficient to ameliorate or prevent sepsis in severe acute pancreatitis. According to Marshall review on PAF inhibitors and Lexipafant study has shown reduced development of organ disfunction in Phase II trial in patients with severe pancreatitis but didn’t show benefit in the Phase III trial .

Conclusion: The first step of treatening sepsis is rightful recognition and identification of septic patient. Non-antibiotic treatment methods are still not investigated enough and present new challenge of modern strategy of sepsis. One of it is Lexipafant, a PAF antagonist, which was presented as a potentionally good trial, but in the end it didn't show significant reduction in mortality of septic patients. Consideration should be given to the fact that a large number of studies have not been conducted on this topic, therefore potential further research should be an option.

Keywords: lexipafant, sepsis, PAF antagonist

Prevalence of placental pathology in preterm births of very low birth weight infants (VLBW)

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Introduction: Very low birth weight (VLBW) infants are classified as infants born under <1,500 g. The multifactorial ethiology of premature birth includes a range of factors which can be broadly classified as: 1) maternal, 2) fetal, and 3) factors affecting the placenta. VLBW infants suffer from various age-specific comorbidities, as a result of the underdevlopement of virtually every organ system. This study aims to determine placental pathology as possible predictor of preterm birth and intrautering growth restriction.

Materials and methods: This research was designed as a retrospective study on VLBW infants born between 2014 and 2019.

Results: A total of 268 patients, with a mean birth weight of 1081±340 g, and a median APGAR score of 8/8, VLBW infants were included in the study. The overall prevalence of placental diseases was 9.7% with a 95% C.I. 6.43% to 13.89%. 1.87% (n=5) of placental diseases were classified as placental abruption, 0.75% (n=2) as imminent placental abruption, 6.34% (n=17) as partial abruption, and 0.75% (n=2) as marginal abruption.

Conclusion: Although the estimated overall prevalence of placental diseases in VLBW births is just bellow 10%, obstetricians and neonatologist should pay attention, due to the possible lethal outcomes associated with placental diseases.

Keywords: placental, preterm births, infants

Review of importance specific biomarkers in laboratory medicine for patients who suffer from rheumatic arthritis

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Introduction: Rheumatic arthritis (RA) is the most prominent systemic, chronic, progressive, the autoimmune rheumatic disease in which the immune system attacks cartilage tissue and bones, and sometimes internal organs. Frequent inflammatory rheumatic disease is present in all ethnic groups. It is more common in women than in men. Data from the literature indicate an increased risk of developing RA in smokers, and environmental factors, viral infections, and genetic factors contribute to the development of this multifactorial disease. Characteristic inflammation leads to joint damage with various pathogenic mechanisms leading to clinical presentation and symptoms.

Review: The review work was designed by searching the database by keywords and professional literature (Pub Med and IntechOpen. Based on the concentration of anti-carbamylate protein, it can be predicted how much the patient's joints are affected by the autoimmune disease. The diagnosis of rheumatic arthritis is made based on history, clinical examination of the patient, radiological and laboratory examinations (leukocytes, SE, CRP, Reuma factor, anti-CCP). The results showed that MMP (matrix metalloproteinase) and YKL-40 (heparin glycoprotein and chitin) are important markers for predicting joint failure. MMP3 is significantly associated with disease activity, inflammatory mediators and cartilage breakdown, and is a potential biomarker of disease severity. The sensitivity of determination of anti-carbamylated protein is thought to be less than ACPA, but simultaneous determination is useful in identification. YKL-40 reflects the current local and systemic inflammation of RA. It is considered an information proinflammatory biomarker. Immunocytochemical study found that YKL-40 is present in polymorphonuclear cells in the synovial fluid of RA patients.

Conclusion: The big correlation is that biomarkers will effectively and safely achieve results on the characteristics of the patient's disease and the treatment group being tested. By analyzing posttranslational biomarkers in RA, one can stratify the patient population in different subgroups showing different results and responses to specific treatments. The future tends to characterize individual profiles, therefore, with the help of genomics and proteomics, significant improvements are expected at the time of diagnosis and response to therapy. At the onset of the disease, additional benefit in the diagnosis of RA is provided by serological markings in addition to ultrasound.

Keywords: RA (rheumatoid arthritis), biomarkers, YKL-40

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ORAL PRESENTATIONS

„MODERN-DAY GENETICS AND ITS FUTURE IN PERSONALIZED MEDICINE"

13th – 14th February 2020

J.J.Strossmayer University of Osijek
Faculty of Medicine Osijek

ORAL PRESENTATIONS

Batičić Lara → Alterations of Neuropeptide Levels among the Gut-brain Axis in Experimental Colitis and the Impact of Dipeptidyl Peptidase IV Deficiency

Brkić Nikolina → Hereditary thrombophilia in pregnant women examined at Policlinic for clotting disorders at General Hospital Vinkovci

Bukulin Dolores → Neuroplastin and ganglioside expression in human amygdala and hippocampus in temporal epilepsy

Laslo Dorian → The comparison of east Croatia autosomal STR frequencies with general Croatian and neighbouring populations

Lončar Ana → EGFR, ALK, ROS1 and PD-L1 molecular testing in patients with non-small cell lung cancer

Radanović Šuk Danijela → Comparative RNA-sequencing analysis of the human thyroid tissue in multinodular goiter

Radanović Šuk Danijela → Global transcriptomic analysis of the thyroid tissue in Hashimoto's thyroiditis

Vučemilović-Jurić Valentina → Role of progesterone receptor gene single nucleotide polymorphism in preterm birth

Zagorac Irena → Immunocytochemical identification of carcinomas of unknown primary in serous effusions

Alterations of Neuropeptide Levels among the Gut-brain Axis in Experimental Colitis and the Impact of Dipeptidyl Peptidase IV Deficiency

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Introduction: Peptidases are considered to exert a crucial role in the etiopathology of inflammatory bowel diseases (IBD, including Crohn's disease and ulcerative colitis). Increasing scientific evidence confirms a causal connection between the central and enteric nervous system where peptidases play a fundamental role in maintaining the homeostasis of their biologically important substrates. Dipeptidyl-peptidase IV (DPP IV/CD26) is an intrinsic membrane-bound and soluble glycoprotein showing a pleiotropic role in the organism. We hypothesized that DPP IV/CD26 contributes to IBD pathogenesis by influencing circulating and tissue levels of neuropeptides among the gut-brain axis.

Material and methods: A trinitrobenzenesulfonic acid (TNBS)-induced (Crohn-like, chemically induced) model of colitis has been induced in CD26 deficient and wild type (C57BL/6) mice. Control animals received the same volume of 50% ethanol and saline solution. Animals were monitored daily for evaluating the disease activity index and were sacrificed 2, 7, 10, 15 and 30 days after TNBS application, representing the whole range of colitis development and tissue healing. Histomorphometrical and pathohistological analyses of colon tissues were performed. Neuropeptides concentrations and protein expressions as well as DPP IV/CD26 enzymatic activity among the gut-brain axis have been determined at both systemic and local levels by ELISA and Western blot techniques.

Results: Our study revealed that CD26 deficient mice constitutionally have significantly ($p<0.05$) higher serum vasoactive intestinal peptide (VIP) concentrations compared to their wild-type counterpairs. VIP concentrations in serum of both mice strains reach their maximum values in the acute phase of colitis. This increase is significantly more accentuated in CD26 deficient mice. Neuropeptide concentrations in colon were increased in both mice strains in acute inflammation as well, with significantly ($p<0.05$) higher values in CD26 deficient mice. Likewise, VIP levels in the brain showed increased concentrations in both mice strains in acute inflammation with significantly ($p<0.05$) higher values in CD26 deficient mice.

Conclusion: The results of our study indicate that mechanisms activated locally in the gut mucosa upon inflammatory events induce changes in neuropeptides in the brain, confirming the importance of the gut-brain axis in IBD. Moreover, DPP IV/CD26 has been confirmed to play an important neuroimmunomodulative role in IBD pathogenesis, which should further be evaluated.

Keywords: neuropeptide, Gut-brain Axis, experimental Colitis

Hereditary thrombophilia in pregnant women examined at Policlinic for clotting disorders at General Hospital Vinkovci

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Introduction: Hereditary thrombophilia is one of the possible causes of recurrent miscarriages and complications in pregnancy. Polymorphisms in genes encoding proteins important for the coagulation system have been recognized as a possible cause of hypercoagulability. Mutations of gene for factor V/Leiden G1691A are associated with adverse events in the second trimester of pregnancy, while mutations of other genes (factor II/prothrombin G20210A, methylenetetrahydrofolate reductase/ MTHFR C677T, PAI-1/plasminogen activator inhibitor) are associated with early pregnancy losses.

Material and methods: This cross-sectional study analyzed the medical records of patients examined at Policlinic for clotting disorders at General Hospital Vinkovci from October 1, 2018 to December 1, 2019, due to pregnancy complications. The analysis included data on age, number of miscarriages and IUGR, presence of mutations in genes for factor V, factor II, MTHFR, PAI-1 and findings that may indicate acquired thrombophilia such as lupus anticoagulant, cardiolipin antibodies, protein C and S. The data were processed with Microsoft Excel statistical tools.

Results: The study analyzed data from a total of 40 patients examined during the observation period. Median age is 31 years, interquartile range from 23 to 41 years. 47.5% of patients (19/40) had 1 or more miscarriages. IUGR was detected in the previous pregnancy in 12.5% of patients (5/40). 12.5% of patients (5/40) were sent for a positive family history of thrombophilia. 95% of patients (38/40) had a mutation in the PAI-1 gene, of which 34% had polymorphism of both alleles. Mutation in the MTHFR gene was found in 55% of patients (22/40), polymorphism of both alleles was present in 32% (7/22). Only 7.5% of patients (3/40) have a factor II gene mutation in heterozygous form, and 5% (2/40) a factor V gene mutation also in heterozygous form. The association between acquired thrombophilia factors and complications in pregnancy has not been analyzed due to incomplete data.

Conclusion: There are numerous studies in the literature that have attempted to establish an association between hereditary thrombophilia and adverse effects in pregnancy. Despite the limitations of our study (small sample of patients, lack of control group), the study found that most patients referred to Policlinic for clotting disorders have mutations in at least one of the genes associated with hereditary thrombophilia. Further studies are needed to demonstrate whether thromboprophylaxis should be administered to patients with hereditary thrombophilia in prothrombogenic conditions such as pregnancy.

Keywords: Hereditary thrombophilia, pregnancy, misscariage

Neuroplastin and ganglioside expression in human amygdala and hippocampus in temporal epilepsy

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Introduction: Neuroplastin (Np) is a transmembrane glycoprotein from the immunoglobulin superfamily. Due to alternative mRNA splicing it occurs in two isoforms one of which, neuroplastin-65 (Np65) is brain specific. Most of the current knowledge about neuroplastin has been obtained from studies in the murine brain, which have shown involvement of neuroplastin in neurite outgrowth, regulation and function of synapses, synaptic plasticity and associative memories formation. However, systematic data on neuroplastin expression and function in human brain are lacking. A few studies of neuroplastin in humans indicate association of Np gene polymorphisms with cognitive functions. Our study also showed changes in neuroplastin expression in human hippocampal samples affected by sporadic Alzheimer's disease and in mouse brain with mutations for familial Alzheimer's disease.

Materials, methods and results: Previous research from our group has shown that the expression and immunopattern of neuroplastin in hippocampus depends on specific brain ganglioside composition. Gangliosides, sialic acid-bearing glycosphingolipids, are ubiquitous cell surface molecular determinants. However, they are predominant in mammalian brain where four complex gangliosides are most abundant: GM1, GD1a, GD1b, and GT1b. Gangliosides have important roles in modulating signal transduction, adhesion and cellular recognition and correct positioning and function of specific membrane proteins. Altered neuroplastin submembrane positioning caused by different membrane ganglioside composition implies a potential interplay between gangliosides and neuroplastin.

Aim of this study was to determine expression of neuroplastin-65 and ganglioside pattern in human amygdala and hippocampal tissue affected by drug-resistant temporal epilepsy.

Amygdala and hippocampal tissue samples were collected with previous patients' informed consent and hospital ethical permission, during hippocampectomy in six patients with drug-resistant temporal epilepsy. Proteins were isolated from tissue homogenates and Np expression was analyzed by Western blotting. We also isolated and analyzed gangliosides from amygdala and hippocampal tissue using high-performance thin-layer chromatography (HPTLC). In addition, we performed immunohistological staining of neuroplastin in control sections of human amygdala. We report altered expression of the Np65 isoform in sclerotic hippocampal tissue compared to surrounding non-sclerotic control hippocampal tissue.

Conclusion: These results show, for the first time, altered Np expression in sclerotic focus of temporal epilepsy in hippocampal tissue. Additionally, we present immunopattern of Np65 in control human amygdala.

Keywords: Neuroplastin, Gangliosides, Epilepsy, Amygdala, Hippocampus

The comparison of east Croatia autosomal STR frequencies with general Croatian and neighbouring populations

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Introduction: We aimed to compare the genetic profile of Eastern Croatian population by studying differences in short tandem repeat (STR) allele frequency between unrelated volunteers from 5 eastern Croatian counties (n=101), general Croatian population (n=195) and four neighbouring countries (Serbia (n=365), Bosnia and Herzegovina (n=1000), Hungary(n=4213) and Montenegro (n=101)).

Materials and methods: Peripheral blood was sampled on FTA cards, and genomic DNA extracted with the use of Chelexmethod. AmpFlSTR® Identifiler Plus kit and ProFlex 3x32 well PCR system were used for the multiplex amplification of 15 STR loci including D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, vWA, TPOX, D18S51, D5S818 and FGA. Allelic variants were identified by electrophoresis on an ABI 310 Genetic Analyser and GeneMapper ID v3.2.1 was used for allele annotation. Allelic frequencies were obtained by direct counting and the exact test of population differentiation wascalculated using Markov-chain algorithm andArlequin v3.5.2.2 software.

Results: In total, 129 STR alleles were detected in the Eastern Croatian population, 8 of which qualifying as rare variants (frequency<0.005). The significant differences between eastern Croats, general Croatian ($p<0.0001$), Serbian ($p<0.0001$) and Montenegrin ($p<0.0001$) population were noticed at D2S1338 STR locus. The CSF1PO allele frequency was in addition, significantly different in Hungarians ($p=0.03684$) and Serbs ($p=0.0306$), while Montenegrins also exhibited different D18S51 ($p<0.0001$) allelic frequency. The STR frequency comparison between east Croatia and Bosniaand Herzegovina, revealed no differences.

Conclusion: Compared to East Croatia, neighbouring populations exhibit different patterns of allelic frequency at 6 examined STR loci, 4 of which determined in comparison to Serbia and Montenegro. Phylogenetic and correspondence analysis should be performed in addition for more detailed characterisation of population relationships.

Keywords: Croatian population, short tandem repeat

EGFR, ALK, ROS1 and PD-L1 molecular testing in patients with non-small cell lung cancer

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Introduction: Lung cancer is one of the leading causes of death in the world. Due to the development of new therapeutic options, the diagnosis of lung cancer involves not only a morphological subtype but also a molecular subtype for patients to receive appropriate therapy. Tumor genomic and/or immunologic biomarker testing now is imperative in the initial assessment and management of advanced non-small cell lung cancer (NSCLC), particularly adenocarcinomas. Molecular testing for epidermal growth factor receptor (EGFR) gene mutations, anaplastic lymphoma receptor tyrosine kinase (ALK) gene rearrangement, ROS proto-oncogene 1 receptor tyrosine kinase (ROS1) gene rearrangements and programmed death-ligand 1 (PD-L1) expression are the current standard in pathology practice for NSCLC. This study aimed to present the EGFR, ALK, ROS1 and PD-L1 mutations in patients with NSCLC lung carcinomas.

Materials and methods: We retrospectively reviewed the 78 NSCLC patient from 1.1.2019 to 30.11.2019. in University hospital Osijek. For molecular testing formalin-fixed paraffin-embedded (FFPE) tissue, cell blocks (CB) or May-Grünwald Giemsa (MGG) stained cytological smears were submitted. The analysis of the tumors for ALK gene rearrangement was performed by immunohistochemistry (IHC) using the Ventana ALK antibody clone D5F3. For EGFR mutational analyses DNA was extracted from formalin-fixed paraffin-embedded (FFPE) tissue or May-Grünwald Giemsa (MGG) stained cytological smears. Samples were tested by polymerase chain reaction (PCR) using the Cobas EGFR Mutation Test, which can detect 41 mutations EGFR gene. ROS1and PD-L1 protein expression detected by IHC analysis using the Ventana D4D6 and SP263 antibody.

Results: The study included 78 unpaired specimens (small biopsies, cell blocks, cytology smears). Fifty-one (35,0 %) participants were female, and the average age was 64 years (range 58 – 73). Ninety-five (65,0 %) participants were male, and median age was 67 years (range 62,75 – 72,00). The majority of testing was performed on small biopsy specimens (33) and cell blocks (32). Cytology smear (13) were tested for EGFR mutations, all negative. The distribution of EGFR mutations according to gender showed a little predominance in the female group (3/5). ALK gene rearrangement is equally presented in both men and women. The prevalence of PD-L1 expression was 24 % (13/54) with a $\geq$ 50 % cutoff and 76 % (41/54) with a $\geq$1 % cutoff.

Conclusions: Patients with advanced NSCLC should undergo mutational testing to evaluate for existing mutations. If such a mutation is discovered, targeted therapy should be considered for first-line treatment.

Keywords: lung cancer, molecular testing

Comparative RNA-sequencing analysis of the human thyroid tissue in multinodular goiter

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2 - Dept. of Nuclear Medicine and Oncology, Faculty of Medicine, Josip Juraj Strossmayer University of Osijek, Osijek, Croatia

Introduction. Nodular goiter is the most common non-neoplastic thyroid lesion, particularly in iodine-deficient settings; however, the transcriptional changes underlying this specific tissue architecture remain unknown.

Material and Methods. V8 release of the Genotype-Tissue Expression (GTEx) project was leveraged to identify and compare 61 nodular goiters to 63 matched control thyroid RNA-sequencing libraries (GRCh38/hg38, STAR v2.5.3a, GENCODE v26). The inverse normal transformed, TMM-normalized read count matrices were corrected for the first three genotyping principal components, the first 60 PEER factors (Probabilistic Estimation of Expression Residuals) and gender (sva R package). The GTEx Histology Image Viewer and sample annotations were used to identify all samples for which nodularity was indicated in the pathologists' notes. Overall, 26054 genes passed quality control. For controls, matching by age/mode of death was used. Pathway analysis (FDR<0.05, generally applicable gene-set enrichment) was performed on gene sets curated from MSigDB v7 (Kyoto Encyclopedia of Genes and Genomes-KEGG, Reactome, Gene Ontology-GO).

Results. The thyroid tissue was sampled from the macroscopically least nodular areas, biasing the results toward the null. As a result, differential expression analysis (limma R) suggested a remarkably similar transcriptome in both normal thyroids and nodular goiters. However, the gene set analysis, which searches for small coordinated expression changes in a pathway, revealed enrichment in modules associated with IRE1-mediated unfolded protein response (GO:adj.P=0.0067), autophagy (GO:P=4.2x10-9), ferroptosis (KEGG:P=0.017), apoptotic signaling pathways in response to endoplasmic reticulum stress (GO:P=0.0066), and wound healing (GO:P=3.5x10-6). Consistent with the recent observations on increased immune cell fractions in nodular goiters, gene sets were enriched for roles in FcγR-mediated phagocytosis (KEGG:P=4.1x10-6), classical antibody-mediated complement activation (P=1.4x10-18), CD22-mediated BCR regulation (P=3.2x10-15), neutrophil degranulation (P=1.4x10-21), signaling by interleukins (P=1.5x10-7), antigen processing (P=7.6x10-6, all P-values: Reactome), and T cell activation (GO:P=8.1x10-7). Finally, a series of enriched modules related to OXPHOS (KEGG:P=3.4x10-12), ribosome biogenesis (KEGG:P=7.3x10-9), citrate cycle (Reactome:P=3.9x10-8), SeCys synthesis (Reactome:P=10-7), PI3K-Akt (KEGG:P=0.0039), mTOR (KEGG:P=0.0073) and MAPK signaling pathways (KEGG:P=0.016) was noted.

Conclusion. We report an exploratory, in-depth RNA-seq analysis of nodular goiters, identifying potentially relevant gene modules.

Keywords: Thyroid tissue, GTEx, nodular goiter

Global transcriptomic analysis of the thyroid tissue in Hashimoto's thyroiditis

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Introduction: In Hashimoto's thyroiditis (HT), autoimmune inflammation results in destructive hypothyroidism. The phenotypic spectrum is highly variable, but the molecular mechanisms that differentially contribute to these phenotypes remain uncharted.

Material and Methods: We analyze bulk RNA-sequencing libraries of the thyroid tissue using v8 data of the Genotype-Tissue Expression (GTEx) project. The GTEx Tissue Image Library was used to identify all post-mortem samples for which HT was indicated in the comments of the GTEx pathologists. In total, 574 samples (37 HT thyroids) and 26054 genes passed the quality control and sample exclusion criteria. We corrected TMM-normalized expression matrices for the first three genotyping principal components (which capture the population structure among GTEx donors), the first 60 PEER factors (Probabilistic Estimation of Expression Residuals), and sex. For controls, 45 libraries were subsetted by matching for age/mode of death. Gene sets (generally applicable gene-set enrichment pathway analysis) were curated from Broad MSigDB v7 (KEGG, Reactome, GO).

Results: Unsupervised computational approaches identified 3 transcriptionally distinct clusters of HT samples, largely corresponding to three distinct pathotypes: focal/mild thyroiditis [n=5, 2208 differentially expressed genes (DEG), limma package, FDR<0.05, fold change >2 vs. controls], moderate HT (n=18, 1221 DEG), and severe/end-stage HT (n=14, 4425 DEG), each dominated by diversity of innate, T and B cell responses. Focal and moderate HT shared enriched gene expression in pathways associated with autophagy (GO:adj.P=1.2x10-5), mitophagy (GO:P=0.02), ferroptosis (KEGG:P=0.034) and wound healing (GO:P=0.031), in addition to alterations in selenoprotein mRNA expression. The emergence of cytokine-cytokine receptor interaction signature (KEGG:P=7.5x10-14), apoptotic (KEGG:P=8.7x10-5) and necroptotic gene signature (KEGG:P=2.3x10-5), cellular senescence (KEGG:P=0.0021) and senescence-associated secretory phenotype (Reactome:P=0.049) marked a transition to moderate and severe HT, together with down-regulation of gene sets associated with cilium assembly (GO:P=1.5x10-6) and the Hippo signaling pathway (Reactome:P=0.01). Finally, the gene modules involved in cell junction organization (GO:P=0.0076), epithelial morphogenesis (GO:P=3.9x10-4), macroautophagy (GO:P=0.0065), mitochondrial biogenesis and metabolism were significantly down-regulated in end-stage HT.

Conclusion: We identify broad transcriptomic divergence across the histological spectrum of HT, and provide a comprehensive atlas of potentially altered pathways in HT.

Keywords: Hashimoto, autoimmune, thyroiditis

Role of progesterone receptor gene single nucleotide polymorphism in preterm birth

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Introduction: Preterm birth is defined as any birth before 37 weeks of gestational age. Based on gestational age, preterm birth is subdivided into three different categories: extremely preterm (< 28 weeks of gestation), very preterm (28 to < 32 weeks of gestation) and moderate or late preterm (32 to < 37 completed weeks of gestation). Approximately 1 in 10 newborns are born preterm and 1 million children die each year due to complications of preterm birth. Preterm birth occurs for a variety of reasons; however, often no cause is identified. The aetiology of preterm birth is multifactorial and includes different pathologies, as well as genetic factors. Better understanding of the causes and mechanisms is the key for development of solutions to prevent preterm birth. One of the genetic factors implicated as a factor for the occurrence of preterm birth is genetic polymorphism in progesterone receptor gene (PGR). The aim of this study is to evaluate whether polymorphism in the progesterone receptor gene both in mother and foetus is associated with susceptibility to preterm birth.

Participants and methods: A total of 100 women with preterm birth and 108 women who delivered at term were genotyped for progesterone receptor gene single nucleotide polymorphism (rs653752) using Taqman assays and real time PCR. Cord blood was collected from their babies (108 at term and 117 preterm infants) and genotyped as well. Possible association between progesterone receptor gene polymorphism and occurrence of preterm birth was examined.

Results: No significant difference in frequency of genotypes between premature babies and those delivered at term was found (Chi-squared test, P<0,05), as well as between women with preterm birth and women who delivered at term (Chi-squared test, P<0,05).

Conclusion: Results of this pilot study suggest that SNP rs653752 in the PGR gene is not a genetic risk factor for preterm delivery. However, further studies of other progesterone receptor gene single nucleotide polymorphisms should be performed in order to evaluate its possible association with preterm birth.

Keywords: progesterone receptor, polymorphism, preterm birth

Immunocytochemical identification of carcinomas of unknown primary in serous effusions

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2 - J.J. Strossmayer University of Osijek, Faculty of Medicine

Introduction and aim of the study: Cytological analysis of effusions is a method by which confirmation is made whether an effusion is of malignant or non-malignant etiology. Metastases from carcinomas of unknown primary in serous effusion are a common clinical problem and immunocytochemistry is applied as an adjunct to the cytological diagnosis. Procedures of acquiring effusions are minimally invasive and cytological analysis is a fast and relatively inexpensive method of diagnostics. The aim of this study is to determine the value of cytological analysis of ascites, pleural and pericardial effusions, in patients with malignant diseases.

Materials and methods: From January 2018 to December 2018, in Clinical hospital center Osijek, the cytological analysis was requested for 89 cases of ascites, 286 of pleural and 10 of pericardial effusion. Spreads were stained by May-Grünwald Giemsa method, and in case of unknown malignant etiology (13 cases of ascites, 37 pleural and 4 pericardial effusion), additional immunocytochemistry was made.

Results: 20 cases, out of 54 that were proven as malignant, were not additionally analyzed, due to the lack of material. Most of the cases that were immunocytochemically analyzed were pleural effusions, 21 cases, followed by ascites, 11, and finally pericardial effusion 2. Mostly used stains were CK7 and CK20, both in 25 cases, BerEP4, 24, and TTF1, 23. Other stains used included: CDX2, CD19, Ca125, ER, ER, RCC, PSA, p40, CK5/6, Napsin A, Calretinin, CD45, EMA and GCDFP. Considering pleural effusions, in 8 cases malignant cells were proven to be originating in lungs, by immunocytochemical stains BerEP4, TT1, and CK7. In addition to those stains, CDX2, CK 19, ER, Ca 125 and CK 20 were most frequently used. In 1 case results yielded breast cancer, and, in the same number of patients, ovarian cancer. The diagnosis was narrowed to lung, breast or genital origin of cancer in 3 cases, while 7 cases were inconclusive. Ascites samples were, in addition to the most common stains, stained with CDX 2 and CK 19 (45% and 55% respectively). Using those stains, 4 cases yielded a tumor of gastrointestinal origin. 1 case was proven to be tumor originating in lungs and 4 cases were additionally stained for Ca 125 and proven to be originating in ovaries. 2 cases were inconclusive.

Both pericardial effusion samples were proven to be positive for malignant cells with tumors originating in the lungs, using stains CK7 and TTF1.

Conclusion: Cytological evaluation of malignant serous effusions, as a fast and inexpensive method with available results in 1-2 days, is a valuable method for diagnosing etiology of effusion and origin of malignant cells.

Keywords: serous effusion, immunocytochemistry, malignancy

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WORKSHOPS

„MODERN-DAY GENETICS AND ITS FUTURE IN PERSONALIZED MEDICINE"

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Faculty of Medicine Osijek

WORKSHOPS

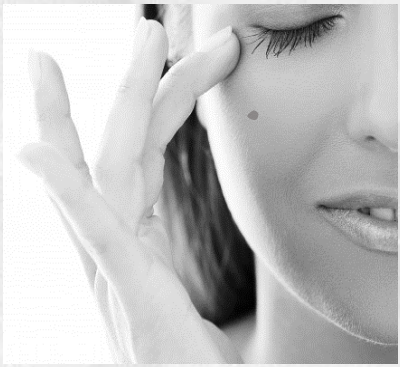
DET. SHERLOCK MOLES

CASE*: WHAT YOU SEE IS WHAT YOU GET... OR NOT?!*

Workshop leader: Marija Šola, MD

Introduction: Identifying moles is not always as easy as it is supposed to be. Mistakes are not allowed, since one man's mistake could mean someone else's end! Discovering melanoma promptly, while it still hasn't invaded and crossed the basal membrane, could be someone's salvation. Melanoma in situ is curable in 100% of cases! Being capable of distinguishing moles from melanomas with, just one quick blink of an eye, isn't possible without dermatoscopy and PHD findings. Therefore dermatologists need to collaborate with other departments (Pathology) in order to get correct results.

Workshop description: Today, we'll try to present you moles and skin lesions similar to them which could, perhaps, mislead you. Later on, we will give you some tips and tricks on how to distinguish them and to become an expert in your job.



INTRO TO SUTURING LIKE A SURGEON

Workshop leaders: Ana Kvolik Pavić, MD; Daniel Čorak; Ivan Bjelousov; Sanja Čubela

Introduction: Treating a cut or other small physical trauma is a very useful skill nowadays considering they are very frequent. During the workshop, you will learn skills that are needed to perform suturing techniques required in most Primary care and urgent care settings. Who can apply? All skill levels!! From a beginner to those advanced individuals that need a refresher course!

Workshop description: To make the model as realistic as possible we will use pig's legs as an example of skin. Each participant will get his injured 'patient' to practice how to address certain wounds.

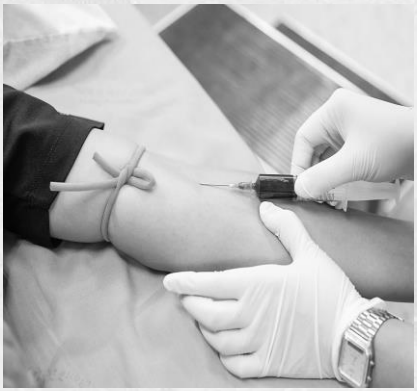
WORKSHOPS

HOW TO BECOME A SUCCESSFUL VAMPIRE: A LESSON TO GO!

Workshop leaders: Ivana Jelavić, Petra Medač, Petar Šušnjara

Introduction: Venipuncture is the collection of blood from a vein which is usually done for laboratory testing. It seems like a simple procedure but... What if something goes wrong? What if you miss the vein? What if you don't collect enough amount of blood or if you mix blood samples and vacutainers? Answers to this and many other questions you will be able to find out on the Venipuncture workshop!

Workshop description: Exercise of venipuncture on the artificial and human hand. Presentation of interesting case reports with the opportunity to solve intriguing problems through a quiz.



HOW TO GET AWAY WITH MURDER

Workshop leaders: Petra Medač, Zvonimir Grgić, Paula Herek, Marija Anđić

Introduction: 23 years old female person has been found dead on Kopika Beach. Her identity is unknown and police have published her photo in the News. Vlatko had recognized the person and immediately called the police. Although he knows that his best friend Marijana, studies medicine in Novi Sad, the dead person looks exactly like her. Even more, he hasn't seen her for 3 days and last time she texted him back was 2 days ago. A few minutes later, Marijana called him. Or was that Marijana?

Workshop description: Groups of 15 students will be analyzing biological samples collected from a dead person, friends, and suspects to determine her identity and her relationship with Vlatko and Marijana. Students will perform DNA extraction from a different kind of samples (blood, saliva smear, hair, toothbrush), and will be introduced with subsequent short tandem repeat (STR) forensic profiling. The workshop will end by comparing the results and establishing relationships between the characters.

WORKSHOPS

KEEP CALM AND TURN THE SIREN ON!

Workshop leaders: Ivan Vilović, mag. med. techn.

Introduction: Have you ever wondered how many lives are saved every day thanks to the professional and well-timed intervention of the emergency medicine team? Or maybe, how many of them do not get saved because of hidden mistakes? Are you interested in how would you react if you were in an ambulance for at least 45 minutes? Delaying such medical assistance, patient's health becomes seriously endangered. That is why it is of extreme importance that the medical staff is capable of quickly estimating a critical situation in which they will, thanks to their knowledge and determination, prevent tragic situations.

Workshop description: For starters, you will have the chance to discover everything one can in din the ambulance vehicle, and how it is adjusted to its purpose. But that is not everything! You will have the chance to perfect auscultation, try out reanimation using a defibrillator, intubation and many other things on our special doll. As a cherry on top, our hosts will give you some secret tips and tricks, which may help you in your future work.

„IT'S NOT LUPUS" DR. HOUSE

Workshop leaders: asst. prof. Silvija Pušeljić, MD, PhD; Višnja Tomac, MD

Introduction: You watch Dr. House and enjoy that intriguing way of diagnosing? What if we told you that you could be part of his team for 45 minutes and you could be the key to resolving the problem? If your answer is yes to both questions then this workshop is the right choice for you. Through the expert guidance of top clinical geneticists, you will learn how to diagnose rare conditions and what makes them think "It's not lupus this time!"

Workshop description: Two clinical geneticist will show you anamnesis and status of their patients with rare disease. After that you will form your „dr.House team" and with their help, try to identify what rare condition lies behind their case. If you go the wrong way they will get you back on the right track. And don't forget: IT'S NOT LUPUS.

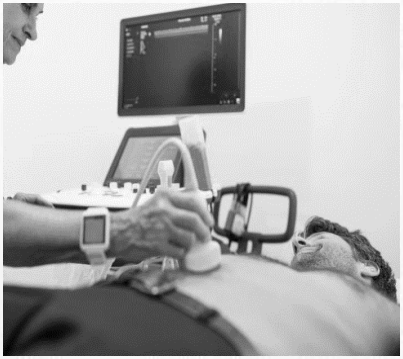
WORKSHOPS

DON'T WAIT, RESUSCITATE!

Workshop leaders: Darjan Kardum, MD

Introduction: Have you seen this 30:2 ratio 100 times and you know everything about it? But what if it's not suitable for everyone? Children are special when it comes to Basic and Advanced Life Support. So if you want to learn how to save a child's life, this is the workshop for you. Learn from a Certified Pediatric ALS specialist how to identify, approach and help a child in the fight for life.

Workshop description: Certified Pediatric ALS specialist will lead you through the basic and advanced skills of life support. You will have the opportunity to test your skills on specially designed dolls for pediatric ALS, so don't miss the opportunity.



YOU CAN SEE INSIDE MY BODY

Workshop leaders: prof. Lada Zibar, MD, PhD

Introduction: Unclear abdominal pain or suspected peritoneal effusion frequently present as urgent clinical challenges. These are some situations in which you could easily respond if you could see inside someone's body. Ultrasound is a noninvasive diagnostic tool that uses sound waves for fast detection of pathological lesions. If you want to learn how to operate an ultrasound machine and recognize the ultrasound appearance of inner body structures, you should definitely visit this workshop.

Workshop description: Experienced nephrologist who applies ultrasound on daily basis will show you the skills how to operate with ultrasound machine and optimize the image. It could help you use the new skills easier in common clinical realities.

WORKSHOPS

ALWAYS BE UNIQUE: MASTER THE STRUCTURE OF CASE REPORT

Workshop leaders: asst. prof. Nina Pereza, MD, PhD; Maja Ploh

Workshop description: The aim of this workshop is to enable participants to learn the structure of case report in the form of scientific article, conference abstract, poster and oral presentation. The workshop is based on different methods of active learning and is an invitation and a sneak preview of the Student scientific summer school organised by the Student section of Medicina Fluminensis, the official scientific journal of the Croatian Medical Association Rijeka and Faculty of medicine in Rijeka.

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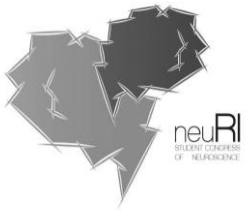
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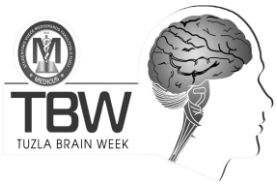
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„THERE'S AS MANY ATOMS IN A SINGLE MOLECULE OF YOUR DNA AS THERE ARE STARS IN THE TYPICAL GALAXY. WE ARE, EACH OF US, A LITTLE UNIVERSE."

- NEIL DEGRASSE TYSON

Josip Juraj Strossmayer University of Osijek
Faculty of Medicine

JOSIP JURAJ STROSSMAYER UNIVERSITY OF OSIJEK