BIOMEDICINE

The Science of the 21st Century

students' symposium

ABSTRACT BOOK

21.05.2022

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BIOMEDICINE
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9:00	OPENING OF REGISTRATION	
9:15 – 10:15	PLENARY LECTURE Prof. Dr Andreas Grabrucker *Is Autism a disorder of abnormal trace metal biology? Insights from in vitro and in vivo models for zinc deficiency.*	
10:15 – 10:45	REGISTRATION	
10:45 – 11:45	OPENING LECTURE **Prof. Alicja Józkowicz** *Bone marrow endothelial cells: choristers or soloists?*	
11:45 – 12:00	Coffee break	
12:00 – 12:35	12:00 – 12:15	**Anna Sowińska-Seidler** *Alterations of the 3D chromatin organization in human limb malformations.*
	12:15 – 12:25	**Katarzyna Ostapińska** *In vivo testing of MBTPS2 mutation found in a patient with IFAP syndrome.*
	12:25 – 12:35	**Krzysztof Jędraszek** *Zebrafish vs quackery around amygdalin.*
12:35 – 13:00	Coffee break	
13:00 – 13:35	13:00 – 13:15	**Ljuba Ponomarev** *SMAD1/5 in lymphatic vessel function and disease.*
	13:15 – 13:25	**Yen Fu Cheng** *Overexpression of human ABCB1 and ABCG2 reduces the susceptibility of cancer cells to the Histone Deacetylase 6-Specific Inhibitor Citarinostat.*
	13:25 – 13:35	**Justyna Derebas** *Caffeine neurotoxicity assessment, used in a chronic system and its combination with pregabalin in behavioral tests in mice.*

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13:35 – 14:25	Coffee break & Lunch	
14:25 – 15:35	14:25 – 14:40	**Anna Sarosiak** *Application of zebrafish model to unravel the involvement of tbc1d24 gene in the development of autosomal dominant hearing loss.*
	14:40 – 14:55	**Monika Maciąg** *Targeting the β-adrenergic receptors to protect against doxorubicin-induced cardiotoxicity.*
	14:55 – 15:05	**Dominika Kresa** *Bioinformatics analysis of genomic structural variants in the development of human genetic disorders.*
	15:05 – 15:15	**Martyna Pociupany** *Biomedical science journey: through oncology, immunology to virology.*
	15:15 – 15:25	**Aneta Malesa** *It's all in the signaling: molecular mechanisms in embryonic development.*
	15:25 – 15:35	**Jakub Książkiewicz** *Life revolves around BMPs.*
15:35 – 16:30	POSTER SESSION AND NETWORKING RECEPTION	
	16:00	Voting for the best presentation and poster
	16:20	Announcement and award presentation ceremony
16:30	CLOSING CEREMONY	

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TABLE OF CONTENTS

ORAL PRESENTATIONS

POSTERS

ORAL PRESENTATIONS

IN VIVO TESTING OF *MBTPS2* MUTATION FOUND IN PATIENT WITH IFAP SYNDROME

Authors: Katarzyna Ostapińska[1], Przemko Tylżanowski[1,2]

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[2] Laboratory for Developmental and Stem Cell Biology, Skeletal Biology and Engineering Research Center, Department of Development and Regeneration, KU Leuven, Leuven, Belgium.

IFAP syndrome is a rare genetic disorder affecting multiple organs in the body and is currently not treatable. To date, several mutations in MBTPS2 (gene responsible for the disorder) have been characterized. Here we describe a novel point mutation occurring in a Polish IFAP patient which results in p.Cys334Tyr change. The mutation results in deficient activity of SITE2 protease, thus negatively affecting cholesterol metabolism. In order to investigate the molecular consequences of this mutation we used an experimental model based on the overexpression of MBTPS2 gene in zebrafish. We misexpressed the mRNA encoding the gene with/without the mutation and carried out comparative morphological analysis. The embryos displayed various developmental defects both in the early as well as later development affecting skeletal system formation. Obtained results highlight the importance of cholesterol in processes underlying i.a. skeletal development.

Key words: IFAP, IFAP syndrome, zebrafish, cartilage, Sonic Hedgehog

ZEBRAFISH VS QUACKERY AROUND AMYGDALIN

Authors: Krzysztof Jędraszek[1], Anna Boguszewska-Czubara[2], Dorota Luchowska-Kocot[2]

[1] Students science club ISOMERS at the Department and Faculty of Medical Chemistry in Lublin, Medical University of Lublin, Poland.
[2] Department and Faculty of Medical Chemistry in Lublin, Medical University of Lublin, Poland.

The incidence of cancer is on the rise due to increased human exposure to toxins. Many pharmacological investigations conducted around the world have proved that many herbal medications contain anti-cancer properties.

Amygdalin (AMY), a glycoside found in common quince and seeds of fruit trees like apricot, cherry, and peach, is one of them. AMY was prohibited and relegated to quackery more than 40 years ago, but thanks to advances in molecular research, we may finally investigate its true anticancer qualities. AMY's impact on cancer growth in vivo utilizing animal embryo models and its molecular action in vitro using cancer cell lines are being studied in particular.

Outcomes of experiments show decreased level of tumor growth, both in cell line and in zebrafish embryo, in opposite to healthy equivalents where AMY exerted no or low toxic effect for viability of cell culture and mortality of fish.

All of the studies in the study lead to the conclusion that zebrafish is an excellent model for carcinoma research. Although AMY is not the greatest example for model studies due to its probable adverse effects and large dose range employed, it is a promising tool for toxicity testing and monitoring of cancer progression in vivo.

Key words: garlic, cancer, S allyl-cystein

OVEREXPRESSION OF HUMAN ABCB1 AND ABCG2 REDUCES THE SUSCEPTIBILITY OF CANCER CELLS TO THE HISTONE DEACETYLASE 6-SPECIFIC INHIBITOR CITARINOSTAT

Author: Yen Fu Chang[1]

[1] Institute of Biomedical Science in Chang Gung University, Taoyuan, Taiwan.

Currently, multidrug resistance (MDR) remains a major obstacle to successful chemotherapy. The overexpression of one of the several ATP-binding cassette (ABC) proteins, such as ABCB1 or ABCG2, in cancer cells can lead to MDR phenotype and poor clinical outcome. Therefore, it is important to understand the pharmacological impact of these drug transporters on the efficacy of chemotherapeutic drugs that are currently in clinical trials. Moreover, studies have shown that the chemosensitivity of these MDR tumors can be significantly restored by directly inhibiting the function overexpression of these drug transporters. Therefore, we utilized the drug repurposing approach to discover therapeutic drugs that can be used to resensitize MDR cancer cells to conventional chemotherapeutic agents. Citarinostat is selective histone deacetylase 6 inhibitor, currently in phase 1bclinical development for multiple myeloma. Our preliminary results indicated that citarinostat is pumped out of cancer cells by ABCB1 and ABCG2.Therefore, we examined the effect of ABCB1 and ABCG2 on the efficacy of citarinostat in cell lines overexpressing ABCB1 or ABCG2.

Key words: multiple-drug resistance, ABC transporter, histone deacetylase inhibitor, citarinostat

CAFFEINE NEUROTOXICITY ASSESSMENT, USED IN A CHRONIC SYSTEM AND ITS COMBINATION WITH PREGABALIN IN BEHAVIORAL TESTS IN MICE

Authors: Justyna Derebas[1], Barbara Miziak[1]

[1] Department of Pathophysiology, Medical University of Lublin, Poland.

Epilepsy is the most common brain disorder in the world. This disease affects as many as 70 million people. Antiepileptic drugs can have adverse effects on cognition, mood and behavior. Treatment of epilepsy focuses on drug therapy. Since 1989, over a period of 27 years (until 2018), 18 new antiepileptic drugs have been introduced to the pharmaceutical market. In total, there were 27 licensed agents for the treatment of epilepsy in 2018. Today, people with epilepsy can expect full seizure control or significant improvement in seizure control. However, in developing countries, due to the lack of specialist knowledge and the lack of adequate treatment options, as many as 80-90% of patients with epilepsy do not receive adequate and effective healthcare. More than 75% of people with active epilepsy are not treated. More than 30% of epilepsy patients do not benefit from the treatment they receive, which translates into their quality of life. The aim of this thesis was to evaluate the adverse effects of pregabalin and caffeine combinations in the chronic system in mice using the funnel test, passive avoidance test and grasp test. The conducted studies did not show any statistical significance of the effect of caffeine on the drug's effect in any of the behavioral tests performed.

Key words: pregabalin, caffeine, mice, epilepsy

BIOINFORMATICS ANALYSIS OF GENOMIC STRUCTURAL VARIANTS IN THE DEVELOPMENT OF HUMAN GENETIC DISORDERS

Authors: Dominika Kresa[1], Adrian Odrzywolski[2], Ewa Hordyjewska-Kowalczyk[3], Przemko Tylżanowski[3,4]

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[4] Laboratory for Developmental and Stem Cell Biology, Department of Development and Regeneration, Skeletal Biology and Engineering Research Centre, University of Leuven, Leuven, Belgium.

Structural variants (SVs) are genomic alterations that span at least 50 base pairs (bp). In the last decades, SVs have been widely studied in terms of the development of human genetic disorders. Whereas numerous algorithms have been developed for SVs detection, such variants discovery remains a challenge due to their complexity and size. There is no single program that can detect all SVs in the genome, thus merging different programs and using additional algorithms should be applied in the SV detection workflows for better results. It is crucial to determine to what extent the detected variants should overlap to consider them as one variant. Computational tests of the wiggle and overlap parameters, which are responsible for the conditioning of merging such variants, showed that the values of 50% for overlap and 1000 bp for wiggle give the highest merging accuracy. The overlap and wiggle parameters with the given values were used for the SVs detection algorithm in the clubfoot family where SVs potentially responsible for inducing the pathogenic phenotype (candidate SVs) were identified. After filtration, 10 candidate SVs were selected, of which duplication within the exon of *LAMB4* was considered the strongest candidate. LAMB4 plays an important role during embryonic development and its disruption can lead to serious consequences resulting in a pathogenic phenotype.

Key words: copy number variations, structural variations, clubfoot, genetics, bioinformatics

BIOMEDICAL SCIENCE JOURNEY: THROUGH ONCOLOGY, IMMUNOLOGY TO VIROLOGY

Author: Martyna Pociupany[1]

[1] Laboratory of Virology and Chemotherapy, Rega Institute, KU Leuven, Belgium.

Introduction: Biomedicine integrates a variety of different scientific fields to bring together new strategies for diagnosis and therapy. Immunological landscape in oncology and microbiology are one of the most relevant disciplines that biomedical research focuses on. My Master Thesis investigated how immunological cell death (ICD) affects immune system, its phagocytic cells and if there is an impact on the immune checkpoints. From that, my research moved onto a PhD in viral oncology. The interactions between oncogenic viruses, their genome and the immunological system is still largely unexplored. My project focuses on post-transplant lymphoproliferative disorder (PTLD), a haemato-oncological disorder driven by Epstein-Barr virus (EBV).

Aim: The goal of this presentation is to familiarize the audience with the scientific projects of biomedical fields.

Materials and methods: For studying ICD, we cultured murine cancer cell lines (LLC and MC38) with chemotherapeutic (paclitaxel) and employed methods like Western Blot, FACS or ELISA. For PTLD project we are working on FFPE biopsies with blood samples and used techniques such as RT-qPCR, exosome, DNA, RNA extraction and MILAN staining.

Results: We were able to characterize that ICD modulates the interface between dying cancer cells and macrophages and we expect to provide an integrated insight into genomic, viral and biological landscape of EBV and PTLD.

Conclusions or Discussion: Through presented projects, it illustrates that the interplay between biomedical fields like immunology, oncology and virology is an area worth investigating that keep pushing the boundaries of scientific discoveries.

Key words: immunology, post-transplant lymphoproliferative disorder, Epstein-Barr virus, viral oncology

IT'S ALL IN THE SIGNALING: MOLECULAR MECHANISMS IN EMBRYONIC DEVELOPMENT

Authors: Aneta Malesa[1], Karol Nowosad[2,3,4], Ewa Hordyjewska-Kowalczyk[3], Przemko Tylżanowski[5], Frederic Lluis Vinas[1]

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[4] The Postgraduate School of Molecular Medicine, Medical University of Warsaw, Poland.
[5] Department of Development and Regeneration, Skeletal Biology and Engineering Research Center, KU Leuven, Belgium.

Introduction: In order to form complex structures from a single fertilized cell, gene expression in the embryo needs to be tightly regulated. One of the main mechanisms of gene regulation are enhancers, cis-acting elements modifying the chromatin structure, which were the focus on my Master thesis in the context of joint development. After that, I decided to focus on early embryogenesis in my research and undertook a project on the role of Wnt pathway modulation in preimplantation mammalian development, with the focus on cell cycle control and cell proliferation in developing blastocysts.

Aim: This presentation will familiarize Biomedical Sciences students with the field of developmental biology and with regulatory mechanisms in early embryogenesis.

Materials and methods: To study the activity of putative enhancers, candidate sequences were cloned using Gateway technology and tested *in vivo* with the use of Zebrafish Enhancer Detection (ZED) assay. To study Wnt pathway activity in early development we currently use mouse embryonic stem cells (mESC) and E2.5 to E4.5 mouse embryos exposed to small molecules, combined with techniques such as IF and confocal imaging, qPCR, FACS, WB, CRISPR technology and RNA-seq.

Results: We were able to show that candidate sequences have regulatory properties in zebrafish larvae. In mammalian development, we have strong indications that Wnt pathway is involved in lineage segregation and potentially influences cell cycle rate and cell proliferation.

Conclusions or Discussion: Strict gene regulation and cooperation of signaling pathways is crucial for proper embryonic development. However, despite their importance, precise signaling networks involved in early embryogenesis are still not fully elucidated.

Key words: developmental biology, gene regulation, stem cell biology, Wnt pathway

LIFE REVOLVES AROUND BMPs

Author: Jakub Książkiewicz[1]

[1] Department of Cardiovascular Sciences, Center for Molecular and Vascular Biology (CMVB), KU Leuven, Belgium.

Introduction: Being a graduate of biomedical sciences has an essential advantage. Thanks to the wide range of subjects learned during the studies, student has a greater chance of experiencing the topic that would trigger his/hers interests. During my Bachelor's and Master's studies I worked on the etiology of hereditary craniofacial malformations – cleft lip/palate and mandibular prognathism. During that time, I became interested in a specific metabolic pathway - Bone Morphogenetic Protein (BMP) signaling pathway. My PhD project focuses on the role of the BMP pathway in lymphatic endothelial cells.

Aim: My presentation aims to show what research projects a graduate student of biomedical sciences can engage in.

Methods: During my Master's studies I've implemented a Whole-Genome Sequencing (WGS) approach, as well as, various molecular biology techniques like PCR, cloning, DNA/RNA isolation to identify potential causative mutations in patients affected by hereditary craniofacial malformations. During my PhD project I aim to utilize scRNAseq, allogeneic transplantation and various immunohistological techniques.

Conclusions or Discussion: My projects showcase just one of many options that graduate student of biomedical sciences. Using WGS we identified a few candidate mutations that can be studied *in vivo* to confirm their pathological influence. Preliminary data of my PhD project shows that LEC-specific BMP depletion in mice results in lymphedema-like phenotype.

Key words: BMP, cleft lip/palate, cell signaling, lymphatic endothelial cells

POSTERS

COVID-19 IN PATIENTS WITH HAEMATOLOGICAL NEOPLASMS

Authors: Hubert Warda[1], Katarzyna Skórka[1]

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Haematological neoplasms are the leading cause of death in children and adolescents throughout the world. Nowadays there is a pandemic of COVID-19 disease which also can lead to death. Unfortunately, patients with haematological neoplasms are at risk of severe COVID-19. This review aims to provide the latest information about COVID-19 in patients with haematological neoplasms including multiple myeloma, acute lymphoblastic leukemia, chronic lymphocytic leukemia and Hodgkin's lymphoma such as fatality rate, risk of severe disease, efficacy of the vaccine against COVID-19 and vaccine-drug interactions.

Key words: COVID-19, haematological neoplasms, CLL, ALL, MM, HL, COVID-19 vaccines

DANIO RERIO MODEL ORGANISM AS A TOOL TO EVALUATE POTENTIAL ANTICANCER PROPERTIES OF YOUNG GARLIC SHOOT EXTRACT IN RHABDOMYOSARCOMA CELL LINE

Authors: Krzysztof Jędraszek[1], Agnieszka Brzezińska[1], Angelika Mastalerczyk[1]

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[2] Department and Faculty of Medical Chemistry in Lublin, Medical University of Lublin, Poland.

The project was carried out under the grant "Students Mini-Grants 2022" concerning the anticancer activity of young garlic shoots.

The project was written thanks to the previous research conducted in SKN ISOMERS at the Department and Faculty of Medical Chemistry in Lublin, which allowed to develop the most effective method of S-allyl cysteine (SAC) and S-allyl mercaptocysteine (SAMC) extraction, as well as to evaluate the possibility of introducing cancer cells into a living animal model of Danio Rerio (Zebrafish).

The project involved laboratory studies based on the use of a rhabdomyosarcoma cell line to compare the amounts of SAC and SAMC in shoot and garlic extracts using HPLC chromatography, measure cellular oxidative stress activity and survival, and examine the effects of SAC and SAMC on the levels of extracellular matrix metalloproteinases (MMPs). The project assessed toxicity in the rhabdomyosarcoma cell line model. Outcomes of study evaluated that SAC is not a main anticancer factor but in combination with other extract’s analytes it can reduce proliferation of cancer cells.

Our studies contribute to the knowledge on natural medicine for cancer, which has to be tested further in pre- and clinical trials.

Key words: young garlic shoots, s-allyl cysteine, cancer

FUNCTIONAL STUDIES OF HEREDITARY HUMAN MUTATIONS LEADING TO CRANIOFACIAL MALFORMATIONS

Authors: Paulina Krzesińska[1], Anna Jaruga[1], Krystian Kuźniarz[2], Przemko Tylżanowski[1,3]

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[3] Laboratory for Developmental and Stem Cell Biology, Department of Development and Regeneration, Skeletal Biology and Engineering Research Centre, University of Leuven, Leuven, Belgium.

Orofacial clefts are the most commonly diagnosed face birth defects during pregnancy. They can be either syndromic or non-syndromic. We can classify facial clefting depending on affected structures: cleft lip (CL), cleft palate (CP), or both cleft lip and palate (CLP). The etiology of CLP is not fully understood, although it has been shown that genetic abnormalities can lead to incorrect palate development. Our research focused on searching for the genetic basis of the hereditary NCLP in a Polish family with three affected members. We found the mutations in *MASP1*, *EHHADH*, and *ACTN2* genes. *MASP1* gene is likely responsible for directing the migration of neural crest cells during embryonic development. Disorders in the *EHHADH* gene are associated with Zellweger syndrome, one of the symptoms of which is a craniofacial deformity. Mutations in the *ACTN2* gene are associated with myopathy and cardiomyopathy. It is probably also related to the occurrence of deep bite defects. Despite the large amount of data on NCLP, the molecular mechanisms of this malformation are not well understood. This knowledge is necessary to understand the consequences of the hereditary non-syndromic occurrence of developmental defects as well as to explore pharmacological solutions to the problem. Our research will generate data that will get us a step closer to understanding the intricate connection between genome, gene transcription, and tissue patterning.

Key words: craniofacial malformations, orofacial clefts, compound inheritance, *EHHADH*, *MASP1*

FUNCTIONAL STUDIES OF MUTATIONS RESPONSIBLE FOR THE HEREDITARY HUMAN LIMB MALFORMATIONS

Authors: Akshaya Ramanujam[1], Ewa Hordyjewska-Kowalczyk[1], Olga Szymaniak[1], Przemko Tylżanowski[1,2], Ewelina Bukowska-Olech[3], Aleksander Jamsheer[3]

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Genetic and epigenetic factors are the basis of many diseases, but the aetiology of many of them is poorly understood. This project aims at the genetic identification of candidate families with hereditary skeletal malformations such as clubfoot (talipes equinovarus, TEV), Pallister-Hall syndrome (PHS), or Greig cephalopolysyndactyly syndrome (GCPS). Next, we will identify the mutations linked to the disorders and carry out functional studies to correlate the genotype and phenotype. Until now, we identified a novel missense mutation c.301C>T p.Arg101Trp in the *TPM2* gene linked to familial isolated TEV. Additionally, we aim to characterize five *GLI3* mutations (c.2255C>G p.Ser752*, c.2017delC p.Gly674Valfs*19, c.2686G>A p.Asp896Asn, c.2721C>G p.Ser907Arg, and c.3018C>A p.Ser1006Arg) causing GCPS or PHS. Thus the next step of our project will examine the consequences of mutation with a gain of functional studies by overexpression of the mutated protein and CRISPR/Cas9 approaches *in vivo* using zebrafish as a model system to demonstrate that they are indeed linked to patient phenotype. Our project results will have implications on the clinical aspects of skeletal development, that may provide a platform to develop pharmacological interventions for some of the inborn skeletal malformations.

Key words: skeletal malformation, idiopathic mutations, *TPM2*, genotype-phenotype correlation, *GLI3*

IN VITRO EVALUATION OF ANTICANCER PROPERTIES OF GERANIOL FOR TWO MALIGNANT MELANOMA CELL LINES

Authors: Katarzyna Pietruszka[1], Paula Wróblewska-Łuczka[2]

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[2] Department of Pathophysiology, Medical University of Lublin, Poland.

Melanoma is the most aggressive type of skin malignance, caused by abnormalities of pigment cells (melanocytes). The tumour progression depends on the risk factors of exposure, including UV exposure, fair skin or hair, or present moles on the skin. Diagnosis is based with the use of “ABCDE” and Glasgow scale, as well as biopsy and histopathologic evaluation. Depending on the result for biopsy, the possibilities to treat melanoma include surgery, chemo- or radiotherapy or novel therapies, i.e. immunotherapy. Due to the major therapeutic challenge, new treatment options have been sought by the researchers to find the effective therapy for melanoma.

Key words: melanoma, geraniol, FM55P, A375

MOLECULAR BASIS OF SKELETAL DEVELOPMENT

Authors: Andrzej Zuzak[1], Michał Bednarz[1], Anna Jaruga[1*], Przemko Tylżanowski[1,2]

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Introduction: Gollop-Wolfgang syndrome is a rare congenital limb anomaly characterized by tibial aplasia and an ectrodactyly. The key objective of this project is to investigate the molecular basis of this disorder. Research focuses on the identification and analysis of mutation in noncoding DNA regions linked to Gollop-Wolfgang syndrome (GWS). Cleft of the lip with or without cleft palate (CL/P) is the most common congenital craniofacial disorder. It can occur as an isolate disorder (CP) or co-exist as a part of a syndrome. Moreover, clefts can be observed as unilateral or bilateral form.

Materials and Methods: DNA from family with hereditary GWS was collected. Methods to identify mutations linked to the phenotypes include Whole Genome Sequencing (WGS), bioinformatics analysis using IGV and PacBio SMRT (Single Molecule Real Time Sequencing). Additionally, we have obtained DNA from another family with GWS. Samples underwent WGS and the data are currently analyzed. The initial part of the CP project focuses on patient genome analysis. We are currently pursuing a candidate gene approach and carrying out Sanger sequencing in all family members available to us.

Results: Linkage analysis revealed a mutation underlying the GWS phenotype to a 5.1 Mb interval at 17pter. WGS and PacBio SMRT determined that mutations described previously in the literature, were not responsible for the GWS in our patients. WGS analysis of new family is currently under way. Following the mutation identification in CP family members, we will focus on demonstrating the link between this mutation and the patient phenotype. To do that we will use a zebrafish-based model system.

Discussion: GWS: Several literature reports identified microduplications within this region possibly underlying the GWS phenotype. In contrast to these studies, our array analysis indicates that in this family this region does not carry microduplications in the region. Identification of genes responsible for NCL/P is extremely complex problem. In spite of the fact that a wide range of candidate genes and risk loci have been associated with clefts formations, clear position regarding the exact mechanisms of inheritance hasn't been still taken. However, IRF6, MAFB, ARHGAP29, 8q24, VAX1 and PAX7 are most often pointed as candidate genes in the literature.

Key words: skeletal development, cleft palate, Gollop-Wolfgang syndrome

NANOPORE SEQUENCING - A BREAKTHROUGH IN MOLECULAR DIAGNOSTICS

Authors: Michalina Pinkosz[1], Paweł Cech[1], Monika Szelest[1], Katarzyna Skórka[1]

[1] Department of Experimental Hematooncology, Medical University of Lublin, Chodźki 1, 20-093 Lublin, Poland.

Nanopore sequencing is one of the newest sequencing methods available. It's based on the flow of ionic current and nucleic acid particles through nanopore structures This technique has been proven to be significantly faster and cheaper than previously used methods. Nanopore sequencing is useful in various industries such as clinical assays or ecology.

Nanopores are naturally occurring proteins that create transmembrane channels. When applying voltage, molecules suspended in the solution start to flow through the nanopores. The disruption of the electric current is detected as a new nucleic acid. Nanopore-based technology allows molecular diagnostics of cancer, including copy number alterations (CNAs), single nucleotide alterations (SNVs) and epigenetic modifications. It may be also used for identification of genetic alterations that qualify patients for targeted therapy. Additionally, sequencing is useful in transplantology because of the Human Leukocyte Antigen (HLA).

Undoubtedly, the biggest advantage of this method is the high speed and simplicity of sample preparation and the sequencing process. Moreover, the equipment based on nanopore technology is small and portable. Nevertheless, nanopore technology has some limitations. These are for example relatively high error rate and low read accuracy. compared with the older sequenators' performance. Despite its issues, nanopore technology is gradually more commonly used in various fields of science, especially medical diagnostics.

Key words: nanopore technology, sequencing

THE ROLE OF MICROBIOTA IN NEOPLASMA DEVELOPMENT

Authors: Maria Cabaj[1], Aleksandra Kamińska[1], Katarzyna Skórka[1]

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The human microbiome consists of a wide range of microbial species. The major abundance of internal microbiota resides in digestive tract. Thus, one of the strongest factors impacting human microbiome is diet. Recent studies prove a relationship between the occurrence of specific microorganisms and cancerogenesis. Microbes can cause neoplastic transformation through genotoxins which increase the chances of host's DNA damage. The occurrence and course of the pancreatic cancer is dependent on microbiome present in various tissues. Contrary to popular belief, even healthy lung tissue is not sterile. Microorganisms present in lungs can lead to cancer development through lung T γδ cells activation. From chemioterapeutic point of view, it is crucial to consider interferences between microbiome and cytostatic medicine for instance gemcitabine or red ginseng extract.

Key words: microbiota, oncogenesis, PDAC, PC, LUAD, genotoxins

THE ROLE OF MIR-196 GENE FAMILY IN DIAGNOSIS AND TREATMENT OF PAEDIATRIC T-CELL ACUTE LYMPHOBLASTIC LEUKAEMIA

Authors: Michalina Pinkosz[1], Monika Lejman[1]

[1] Laboratory of Genetic Diagnostics, Medical University of Lublin, Poland.

Acute lymphoblastic leukaemia (ALL) is one of the most common cancers diagnosed in children, accounting for around 25% of total pediatric cancer cases. T-cell originating ALL (T-ALL) has currently over 90% cure rates, however, the patients have 20% risk of relapse. Presently, immunophenotyping is crucial for T-ALL patient stratification. Analysing a patient's genome shows the molecular origin of the disease, which helps to predict the treatment outcome. In recent years researchers are highlighting the importance of non-coding RNA fractions such as micro-RNA (miRNA) in tumorigenesis. MiRNA are small, endogenous, non-coding RNA particles which regulate mRNA expression. The disturbance of miRNA-mRNA interaction causes errors in the cell cycle regulation and control, resulting in various pathologies such as cancer. MiR-196 is a gene family strongly connected with HOXA genes which are involved in leukemogenesis. The upregulation of miR-196 is present in many cases of paediatric T-ALL. Recent studies show that MiR-196 can be used both as a T-ALL biomarker as well as potential therapeutic target. In this essay we present the origin, function of miR-196 family and discuss their utility in diagnosis and potential therapy of paediatric T-ALL.

Key words: paediatric leukaemia, miRNA, T-ALL, miR196

TOXIC AND NUTRITIONAL OPTIC NEUROPATHIES—REVIEW

Authors: Alicja Forma[1], Joanna Kobak[1], Magdalena Tyczyńska[2]

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Introduction: Optic neuropathies form a group of conditions with various origins and different causing factors.

Aim: The characteristic of both genetic and acquired causes of optic neuropathies. Furthermore, the poster includes a specification of toxic optic neuropathy. Moreover, it indicates the danger and the negative impact of nutritional deficiencies, vitamin absorption disorder and anaemia on our sight.

Materials and methods: Review, based on previously published article: https://www.mdpi.com/1660-4601/19/5/3092?fbclid=IwAR14QgoIG6QXeOd6FY4GIX7dDJaC03PrTvcQyCbfjMlKIxkD9fGGDeFA9Ic

Results: Alcohol is no longer recognized as a toxin itself, however alcoholism primarily leads to deficiency of essential nutrients and vitamins, which appears to be the risk factor of developing nutritional optic neuropathy. Vitamin B12 and folate shortages as well as the cyanide in tobacco may be linked to demyelination of the optic nerve and with free radicals may impair the mitochondrial respiratory cycle with the damage of mitochondrial DNA. Smoking might significantly increase disease penetrance among the carrier's mutation characteristic for Leber hereditary optic neuropathy (LHON). Currently it is believed that term 'alcohol-tobacco optic neuropathy' is misleading, as interaction or synergism of alcohol and tobacco as triggering factors has never been categorically proven.

Conclusions: The therapeutic options in the treatment of optic neuropathy strictly depend on the causative factor. Nutritional deficiencies are treated with appropriate supplementation, so it is vital to truly determine the missing vitamins and elements.

Key words: toxic optic neuropathy, alcohol, tobacco, drugs, nutritional deficiencies