INVITED SPEAKER ABSTRACTS

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Making Biotechnology from Nanostructured Systems

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In this presentation I will explore the design and assembly of nanostructured systems with inspiration from biology. Here we consider the biological inspiration as not the approach typically adopted to construct synthetic materials inspired from nature. In contrast, the approach discussed in this presentation utilizes biological components in the assembly of smart nanostructured systems. The term "biological engineering" is used to describe a discipline that embodies this approach. Indeed, in 1970, this term was introduced formally with the intention to integrate engineering with biological systems to move beyond single disciplinary areas such as medicine, agriculture, or fermentation engineering. In this presentation we further refine the discipline of "biological engineering" to be one that utilizes biological proteins, molecules and lipids in combination with synthetic materials to assemble smart nanostructural systems. I will illustrate this discipline with the assembly of nanostructured systems that are targeted for applications such as diagnostic, therapeutic, biofuel cell, or tissue enhancement/replacement in the body.

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Studies on bioactive peptides with analgesic activity

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The process of inflammation is a part of the immune response. This process is often associated with the onset of pain. Thus, both processes are closely related and have intertwining mechanisms of occurrence and action. Currently, there are many analgesics for pain treatment but unfortunately, they have many undesired secondary effects or are not strong enough to suppress the pain. Thus, the search for new alternatives continues. New tetrapeptide analogues of FELL and bioconjugates with a general formula X-Aaa-Glu-Bbb-Ccc-Z, where X is NH_2 or pyrrole moiety, Aaa = L- or D-Phe, Bbb and Ccc are Nle, Ile or Val and Z = NH_2 or COOH were synthesized using standard solid phase peptide synthesis by means of Fmoc/OtBu strategy. New molecules were designed in order to study the influence of hydrophobicity on the analgesic activity as well as to search a synergic effect between both part of the targeted bioconjugates. Analgesic activity was examinate by the Paw-pressure test (Randall-Selitto test). Some interesting structure-activity relationships were revealed and will be discussed.

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The role of copper and solid lipid nanoparticles in parkinson's disease pathogenesis and therapy

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Parkinson's disease (PD) is a neurodegenerative disorder characterized by the loss of dopaminergic neurons in the substantia nigra pars compacta (SNpc). The disease hallmark is Lewy bodies (LBs) in surviving neurons, primarily composed of α-synuclein protein. In PD, α-synuclein is often phosphorylated at Serine 129 (pS129), promoting its aggregation and interaction with metal ions like copper, which accelerates α-synuclein aggregation into fibrillar plaques. This study explored copper's role in PD pathogenesis, focusing on α-synuclein clearance via autophagy and ubiquitin- proteasome system (UPS). A human neuronal model, SH-SY5Y cells, was developed, demonstrating dopaminergic characteristics suitable for PD studies. Copper overload was induced using copper chloride, which increased autophagy markers and altered the PI3K/AKT/mTOR pathway, promoting autophagy but impairing autophagic flux and UPS, leading to increased α-synuclein levels. The study also examined copper overload in cells overexpressing α-synuclein, revealing that copper exposure increased cytotoxicity and lysosomal activity, further impairing protein degradation mechanisms.

Additionally, solid lipid nanoparticles (SLNs) containing grape seed extract (GSE) and dopamine (DA) were tested, showing cytoprotective effects against rotenone-induced toxicity, oxidative stress, and increased α- synuclein levels. SLNs show therapeutic potential for noninvasive approach to PD, supporting the hypothesis that copper dyshomeostasis compromises α-synuclein clearance, exacerbating cellular dysfunction and stress.

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Bacterial cellulose based biocomposite materials for medical and technical purposes

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Bacterial cellulose (BC) is currently one of the most popular environmentally friendly materials with unique structural and physicochemical properties for obtaining various functional materials for a wide range of applications. So, new BC-based biocomposite materials for medical purposes with antibacterial, regenerative, and hemostatic properties in the form of films, hydrogels, aerogels, and tubes were obtained at the Department of Biotechnology, biochemistry and bioengineering of National Research Ogarev Mordovia State University. Also, new biocomposites with a high sorption capacity for fluorine ions and aerogels with a low coefficient of thermal conductivity, high sound absorption coefficients, and high mechanical strength has been developed. The results prove the potential of BC-based biocomposite materials for biomedicine, environmental monitoring, and for various industries.

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Keywords: biocomposite materials, bacterial cellulose, biomedicine.

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Oil Production by Microalgae; Importance of Native Strains

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Microalgae are considered as an interesting sustainable source of high- and low-value oils. In this regards choice of right strain(s) is of importance. This paper reports the major findings of our extensive research on oil production by indigenous Dunalilla strains. More than 130 strains were evaluated in terms of oil production. Among them five isolates of D. sp. ABRIINW-B1, -G2/1 and -I1, -CH_2, and -SH33 were selected. The oil content of the strains reached to 42, 36, 47, 21, and 33% of dry matter, respectively. Fatty acid (FA) profile analysis of the strains showed that all of them are suitable sources for polyunsaturated fatty acids. But the strain D. sp. ABRIINW-I1 is the only one enable to produce remarkable amount of docosahexaenoic acid (DHA). Then, the strain was screened out for the subsequent lipid examinations. Results showed that it could produce the maximal DHA of 36 % total FA by volumetric production of 30 mg L^{-1} in 0.5 M, 25 °C, 1200 µmol photon m^{-2} s^{-1}. Expose of this strain to CO_2 led to changes on the FA profile in a way that production of DHA tended to zero and the overall FA profile became suitable for biodiesel production. On the other hand, D. sp. ABRIINW-CH_2, and -SH33 maintained the ability to produce oil with suitable nutritional value even at high CO_2 concentrations up to 30%. Further studies showed that although these two strains had a same reaction to high CO_2 concentrations but they used different molecular pathways for this response. These findings show the importance of studies on native microalgal strains for optimal oil production.

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Additional open reading frames in the annotated mRNAs of mammals and plants may encode unknown proteins

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Although over 25 years have passed since the first eukaryotic genomes were introduced, the questions "How many genes does a genome have?" and "How many different proteins are synthesized in a cell?" are unanswered. The initiation points for the synthesis of most eukaryotic proteins are located at the beginning of the mRNAs (CAP) and determined by a CAP-dependent scanning mechanism. However, due to the Internal Ribosome Entry Sites, translation can also begin at points of mRNAs far from the CAP. In this work, all annotated mRNA sequences from human, rhesus monkey, chimpanzee, mouse, rat, rice, corn, soybean, black alfalfa, black poplar, tomato and wine grape species were searched for additional open reading frames (ORFs) that could encode polypeptides at least 200 amino acids long. At least, one unknown ORF/polypeptide was detected in some mRNAs of these species. Human, rhesus monkey, chimpanzee, mouse, rice, and maize mRNAs appear to be more abundant with additional ORFs. Comparison of those putative proteins with proteins from the SwissProt and NCBI RefSeq resources showed that the more than 90% of “novel” proteins were not previously annotated. These results indicate

that the same mRNA can encode more than one protein, and this phenomenon is widespread.

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Nutrigenomic in Sparus aurata fed with polyphenols, sustainable by-products of olive oil production process: preliminary data

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The aim of this work was to apply the concept of circular economy for sustainable fish feeds production. In Sparus aurata the effect of diets integrated with polyphenols purified from olive mill wastewater by-products was evaluated on gene expression level (nutrigenomic approach). Farmed gilthead seabream were categorised into two groups: one control group -ctrl- (9 specimens in 3 tanks) fed with standard feed, the other group (9 specimens in 3 tanks) treated -tr- with conventional feed supplemented with polyphenolic extracts. Post-mortem, a portion of each posterior intestinal (PI) district was sampled, from which mRNA was extracted to conduct novel RT-qPCR assays. For FABP2 (fatty acid binding protein2), involved in metabolic pathway, it was observed a trend of down-regulation in tr group; for SOD1 (superoxide-dismutase1) enzyme characterised by a 'ROS scavenger' role, an over expression in ctrl group was registered; for the pro-inflammatory interleukin, IL-12, a significant (p-value<0.01) down-regulation of expression was detected in tr fish. The outcomes revealed anti-inflammatory, anti-oxidant properties of polyphenols such as their function in reduction of synthesis, uptake and transport of fatty acids.

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Development of genetic biotechnologies at the national academy of sciences of belarus

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Genetic researches at the National Academy of Sciences of Belarus are carried out and successfully implemented in the practice of agriculture and forestry, healthcare, sports, forensics and environmental protection. In the field of agriculture, DNA markers have been developed for marker-assisted selection (MAS) of 15 crops, MAS technology has been introduced for domestic pigs and cattle. In the field of forestry, an assessment of genetic resources of the main forest-forming species of Belarus was carried out, molecular genetic methods for identification of diseases and pests of forest woody plants were developed. In the field of human genetics, testing techniques of predisposition to a number of diseases have been developed and improved, and more than 24 thousand people have received genetic passports. Genetic testing programs have been developed for sport to select and profile promising athletes, assess the risk of pathologies. Research is being conducted in the field of pharmacogenetics and genetics of longevity, microbiome genetics. In the field of environmental protection, DNA barcoding methods are being developed for the genetic identification of rare and endangered species of plants and animals, as well as invasive species. Innovative structures have been created, as well as biobanks for storing genetic material.

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Evaluation of microbial diversity at water ecosystems as a reflection of tolerance to toxicants

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Microbial biomass and composition in saline coastal lakes are prone to fluctuations in hydrological regime, geological characteristics, climate conditions, and human interventions, which by all means produce a complex network of factors to be considered. CARD- FISH, DAPI staining, chemotaxonomy, sequencing of 16S-rDNA gene fragments were used to evaluate the microbiota abundance and diversity in water and sediment samples from Lake Butrinti, Albania. Results hardly explained by the physical-chemical characteristics alone, or by the correlations with other influencing factors, were analyzed based on the toxicity levels as evaluated through modified luminescence of Vibrio fischeri (ISO 11348-3-2007). In conclusion, differences in microbial taxa present in waters versus sediments of the same sampling stations can be interpreted as a reflection of the chemical-physical characteristics of the environment (i.e. oxic vs anoxic), or related to the vicinity to coastal settlements, and/or as plasticity to adapt to toxicity of waters measured straightforward via single-cell sensors.

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Dark side of nanotecnologies

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Globalization represents the global integration of international trade, investment, information technology and cultures. A global programmer for sustainable future until 2030 was adopted by UN. It includes 17 development goals with regards to climate, social and economic problems. Analysis of these goals,

demonstrates that, the environmental safety appears to be the key condition for achieving all 17 SDG. Yet, now, about 40% of serious diseases and untimely deaths are connected with harmful environmental effects. In 150 years, the speed of introduction of critical technologies has decreased from 50 to 6 years. That makes the necessity of protentional hazards are of vital importance. Nanotechnology carries a significant impact and serves as a revolutionary and beneficial technology, across various industrial domains. It includes communication, medicine, transportation, agriculture, energy, materials, manufacturing and consumer products. Fast growing market of nanotechnologies and nanoproducts ($1.76 billion in 2020, and is projected to reach $33.63 billion by 2030) is expected to improve our life and make it safer. But there remain many unknown details about the interaction of nanoparticles and biological systems. Therefore, more information on the response of living organisms is necessary. The presence of nanoparticles of varying size, shape, chemical composition and surface characteristics is needed to understand and categorize the potential toxic effects. The key questions that arised are the following: Can nanoparticles interact with living organisms? Which characteristics of nanoparticles are relevant for health effects? What are the health implications of nanoparticles used in biomedicine? How should harmful effects of nanoparticles be assessed? What are the effects of nanoparticles on the environment? Among the widely used engineering nanomaterials, metal nanoparticles are extensively applied in biomedicine, yet, the studies on the hemolytic effects and metabolic disorders of them in human erythrocytes are still limited. Erythrocytes are the dominant cell type (90 %) in blood, and the hemolytic effects of nanoparticles via direct interaction with erythrocytes must be the object of intensive study. In our work we have shown dose-dependent oxidative damage and energy metabolism disorder contributed to the hemolytic effects of AgNPs in vitro. To estimate the hazard of actual levels of nanoparticles for human cells and their structures as well as mechanisms and the rate at which nanoparticles penetrate into the human body tissues intensive investigations must be conducted before the rollout of a new products on the nanomarket.

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Red yeast cell factory – from the laboratory to industrial production

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Carotenogenic yeasts are present in various natural environments and in many foods and food materials as well. Moreover, they are widely used in biotechnology, predominantly as producers of pigments and single cell oils, rarely as a source of enriched biomass. Microbial food cultures have come under various regulatory frameworks during the last decades. To an updated inventory of microorganisms covering a wide range of foods some oleaginous red yeasts were included. In previous study several strains of pigmented yeasts newly recognized as "safe" were evaluated regarding their biological effects. Present study is focused on complex characterization and large-scale production of enriched red yeast biomass using the yeast strain Rhodotorula toruloides. Optimization of production parameters and their influence on composition of yeast biomass in laboratory conditions is evaluated. Further, under cooperation with industrial partner Algae Farm, Ltd., technology transfer to pilot-plant production of *R.toruloides* biomass in fed-batch 2000 L fermentor is discussed. Processing of yeast biomass and characterization of its complex composition, safety and biological effects are introduced as well. Finally, based on approval of Czech legal authorities, industrially processes *R. toruloides* biomass was brought out to market as a food supplement marketed under the commercial name LivelinR. This supplement contains processed biomass with the optimum content of valuable metabolites and considerable biological activity, which can be recommended for daily intake to prevent diseases connected with oxidative stress, many metabolic disorders, immunity disbalance and biological senescence.

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The fluoroquinolone antibacterial drugs are very powerful bleaching agents of the chloroplasts in the flagellate euglena gracilis

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Fluoroquinolones are a class of synthetic antibacterial agents containing a fluorine atom at the C-6 position, increasing their antibacterial activity and pharmacokinetic properties. Since chloroplasts are of bacterial origin fluoroquinolones have been found to have strong efect on the chloroplast system of the photoautotrohic flagellate *Euglena gracilis* manifested as a permanent bleaching connected with the the loss of photosynthetic capability and the damage of chloroplasts and their genomes. Loss of part or the entire plastid genome without loss of nuclear genes makes *E. gracilis* an attractive model for studying reductive plastid evolution. Fluoroquinolones are classified based on the spectrum of their antibacterial activity into four generations. We have studied the effect of selected fluoroquinolone derivatives from all those groups on the chloroplasts of *E. gracilis* assessing their ability to induce white mutants depending on the various concentrations of the drugs and in three time intervals (24hours, 7 days and 14 days). The most effective bleaching agent was moxifloxacin. Insights from the study of fluoroquinolones on the chloroplasts of *E. gracilis* may bring new information on genetics and evolution of organelles and their possible application in biomedicine and biotechnology.

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Key words: *Euglena gracilis*, chloroplasts, fluoroquinolones, bleaching, antibacterial agents

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Plant biostimulants: Tools to enhance crop tolerance to abiotic stress in a climate change scenario

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Plant biostimulants have emerged as promising agronomic tools, complementary to classical breeding and genetic transformation or genome editing approaches, to enhance the responses of crops to abiotic stresses such as drought and salinity. These adverse environmental conditions are increasingly prevalent due to climate change effects, particularly in arid and semi-arid regions, and threaten food security worldwide since all our major crops are sensitive to abiotic stress. Biostimulants, which include a diverse collection of inorganic elements, natural and synthetic organic compounds, biological extracts, and beneficial microorganisms, are defined by their positive effects on plant growth, crop yields and tolerance to environmental stressors. Balox® is a commercial biostimulant of plant origin, made from rice and oat husk protein hydrolysates and containing polyphenols and glycine betaine as additional active components. In this talk, we will show the effects of Balox® application on tomato and lettuce plants, directly protecting the root absorption zone in the presence of salt, stimulating growth and increasing photosynthetic pigment contents, reducing the accumulation of toxic ions (Na^+, Cl^-) in roots and leaves while increasing the concentration of 'beneficial' cations (K^+, Ca^{2+}), and lowering the level of salt-induced oxidative stress in the plants.

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Bioprocess intensification and the model-based design

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Recent advancements in microprocess engineering highlight the significant benefits of microreactor technology in improving product yield, purity, and process efficiency compared to traditional bulk reactions [1]. Innovations such as continuous processing, flow chemistry, high-throughput screening, and process intensification are reshaping process design and engineering. Despite the potential of microreactors to revolutionize (bio)chemical synthesis, there is a lack of detailed proposals for replacing existing industrial processes. However, recent developments in systematic approaches and tools aim to bridge the gap between research and industry, facilitating the transfer of lab-on-a-chip technologies to industrial settings. This work explores microscale (bio)process development using scale-up/numbering-up concepts and modeling-based optimization. It emphasizes the importance of understanding fluid dynamics at the microscale and its impact on transport phenomena. Theoretical insights into transport phenomena and kinetics are illustrated through model validation for biocatalytic processes in the packed bed microreactor "between two-plates[2,3].

References

1. Pohar, A., and Plazl, I. Process Intensification through Microreactor Application, Chem. Biochem. Eng. **2009**, Q.23(4) 537–544.
2. R. Ambrožič, U. Krühne, I. Plazl. Microfluidics with redox-responsive hydrogels for on-demand BPA degradation. Chem. Eng. J., **2024**, 485, 149542, doi: 10.1016/j.cej.2024.149542
3. T. Menegatti, I. Plazl, P. Žnidaršič-Plazl. Model-based design of continuous biotransformation in a microscale bioreactor with yeast cells immobilized in a hydrogel film. Chem. Eng. J., **2024**, 483, 149317, doi: 10.1016/j.cej.2024.149317.

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Development of sustainable biocatalytic processes for active pharmaceutical ingredients production

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Biocatalytic processes offer transition to more sustainable manufacturing of pharmaceuticals, especially when starting from the bio- based resources. Reactor miniaturization, continuous operation, and integration with in situ product removal, process analytics, and cascade reactions that reduce the number of process steps enable process intensification. Examples of efficient biocatalytic processes development comprising enzyme and substrate screening, medium engineering, and biocatalyst immobilization to intensify biocatalytic processes will be presented.

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Importance of microbes in circular bioeconomy

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Microorganisms are key to a functioning circular economy where natural resources, including biological waste, are converted by microbial cells into added value products (e.g. biogas, bioethanol, biodegradable polymers, compost) utilized by so-

ciety. Microbial polymers have an enormous potential as they can be produced from renewable biogenic resources (waste) under well controlled conditions. This approach can help address growing concerns about the depletion of natural resources caused by an increasing human population, waste production, and accumulation.

Over the past few decades many biopolymers originating from various types of microorganisms have been reported. This presentation will discuss an integrated biotechnological/chemical process, which allows the transformation of agricultural and other biorefinery by-products into value-added biodegradable polymeric materials which may be used in the areas of agriculture, cosmetics, medicine, as well as in household products, coating systems and as additives to compostable plastic packages of extended shelf life.

This presentation will also describe the biosynthesis and production of biodegradable microbial polymers, including polyhydroxyalkanoates (PHA), bacterial cellulose (BC) and poly-gamma glutamic acid (PgGA), their most important industrial applications and ecological advantages.

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Applications of CRISPR technology in development of transgenic mice for modelling rare genetic disorders and beyond

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Research on rare diseases with low prevalence, often suffers from a lack of sufficient patient samples, hindering comprehensive exploration of symptom pathogenesis. Particularly, rare genetic diseases that manifest neurodevelopmental phenotypes pose unique challenges for both diagnosis and therapeutic intervention. These disorders are frequently driven by loss of function mutations in specific genes, and exhibit a wide spectrum of clinical manifestations, making their study and treatment particularly complex. Genetically engineered mouse models aka transgenic mice are indispensable tools to study the underlying mechanisms of human diseases, conduct preclinical research, and develop potential therapeutic strategies. Our platform develops genetically engineered mouse models by CRISPR genome editing in mouse embryos. We have created a multitude of knockin/knockout mouse models for rare genetic diseases as well as for fundamental biomedical research. I will explain our approach in CRISPR genome editing and subsequent phenotypic characterization of mutant mice, and discuss two of the models we have developed. One of these models has been genetically humanized to mimic Rahman syndrome, an epigenetic rare disease, whereas the other carries multiple knockout mutations to allow humanization of its liver by repopulation with human hepatocytes.

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Malignancy Assessment of Colorectal Polyps Using AI

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The application of computer-aided diagnostics (CAD) systems has received considerable attention in recent years due to their capacity to contribute to the high accuracy and efficiency of medical diagnoses. These systems can contribute significantly to the pathological diagnosis of colon polyps in medical imaging, thus contributing to the prevention and treatment of colorectal cancer. This study introduces a comprehensive dataset of colon polyps. The dataset comprises 201 patients, 202 colonoscopy videos total 422 minutes and 2.14 (±4.35) minutes per patient and 1903 microscopy images collected prospectively from the Kayseri City Hospital between 2019 and 2021. The dataset comprises colonoscopy and microscopy images with their pathological diagnosis result for 399 polyps diagnosed pathologically. In this data set, results obtained by staining proteins such as p53 (clone: bp53-11), Ki-67 (clone: 30-9), CD34 (clone: QBend/10), PD-L1 (clone: SP142), BRAF (clone: V600E) and VEGF (clone: SP125) using the immunohistochemistry (IHC) method are also available. This talk will present statistical analysis and interpretation of this multi-modal dataset, findings from the machine and deep learning models to achieve automation and uniformity of polyp and cancer diagnoses using colonoscopy and histopathology images.

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Growth differentiation factor 15 levels in eating disorders patients

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Growth differentiation factor 15 (GDF15), a stress-responsive TGF-β superfamily cytokine, is increasingly recognized biomarker for regulation of metabolism, energy homeostasis, and body weight but few studies have investigated the connection between GDF15 levels and malnutrition whether by excess or by deficiency in patients with eating disorders (EDs). In humans circulating GDF-15 change with age, smoking, drug abuse, pregnancy and metabolic stresses such as adiposity, starvation, mitochondrial dysfunction, diabetes and cancer. The broad normal range in human under 40 years-old is 200-1200 pg/mL. In this scenario, we aimed to investigate GDF-15 role measuring its serum concentration in EDs patients because is still poorly understood. Serum GDF15 concentrations were evaluated using a commercially available ELISA kit in 320 EDs patients (divided in five major groups as described in DMS-V) and 80 healthy controls. No statistical differences were observed in Anorexia Nervosa patients (both restrictive and purging su-

b-types) vs healthy control group matching by age and sex. On the contrary Binge Eating patients showed high levels of circulating GDF-15 probably due to high body max index and adipose tissue. Moreover GDF-15 was correlated with some critical blood values such as hematocrit, red blood cells and cholesterol suggesting once again its possible therapeutic applications.

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Lysosomal Enzymes in Parkinson's Disease

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Lysosomal enzymes have been linked to neurodegenerative diseases, including Parkinson's disease (PD). The main function of these enzymes is the degradation of intracellular and extracellular macromolecules. Mutations in the genes coding for lysosomal hydrolases cause lysosomal storage diseases and to date about seventy diseases have been described. Several lysosomal storage diseases exhibit neurodegeneration in the central nervous system as one of their main features. PD is the second most common neurodegenerative disorder and the most common movement disorder. Several genes have been associated with PD, in particular mutations on gene encoding for lysosomal enzyme beta- glucocerebrosidase is considered one of the major risk factors for PD. More recently GWAS have also identified additional candidate genes encoding for lysosomal proteins such as CTBS, GALC, GUSB, NEU1, CTSD, ASAH1, LAMP1, ATP13A2 as risk factors for PD. Altogether these findings show a strong contribution of lysosomal dysfunction to influence the development of PD. The important role that the lysosome seems to play in PD.

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Non-Invasive and Real-Time Approach to Detect Osteoarthritis Using Machine Learning Algorithms

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Osteoarthritis is a degenerative joint disease that is common worldwide. The disease is classified differently as early and late stage. In late stage patients; knee pain, limited range of motion and joint inflammation are the reasons for clinical admission. Nowadays, in addition to clinical examination, osteoarthritis is diagnosed with methods such as MRI, X-ray and Arthroplasty. The fact that these diagnostics are long, expensive, cause radiation exposure and are not reproducible due to invasive methods creates the necessity for new diagnostic methods. The study aimed to propose a classification diagnostic method by acquiring vibroarthographic signals and temperature data to overcome existing methods' limitations and investigate this method's effectiveness. In this regard, data were collected from 16 patients diagnosed by their clinician for the presence and stage of knee osteoarthritis using an electro stethoscope and a thermal measurement system produced during the study, over a period of approximately 5 minutes. The collected signals were first preprocessed and then processed by feature extraction. The extracted features were eliminated by Neighborhood Component Analysis. Afterwards, the extracted features were evaluated with machine learning models, it was determined that the most suitable model for classification was SVM Poly with 92% accuracy.

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Food fortification with vitamin D and risk of cardiovascular diseases

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Vitamin D (VD) deficiency is known to contribute to the development and/or outcomes of cardiovascular diseases such as atherosclerosis, coronary artery disease, heart failure and aortic stenosis. The active metabolite of VD, 1,25-dihydroxycholecalciferol, acts by binding to the nuclear VD receptors (VDR) and affecting the expression of almost 3% of the human genome. In the cardiovascular system VDRs are present in fibroblasts, vascular endothelial smooth muscle cells and cardiomyocytes. The production of the active form of VD is stimulated by VDR activation and may lead to upregulation of angiotensin-converting enzyme-2, enhance renin production, control the expression of metalloproteinases that are responsible for the induction of heart failure, regulation of plaque formation and the development of atrial fibrillation through various mechanisms. Overall, there is evidence that VD plays roles in the regulation, physiology and pathophysiology of diseases and the cardiovascular system. VD deficiency can affect the sensitive balance of this system leading to the development of disease. VD deficiency is so common that it has been characterized as a global pandemic. Reasons for this are decreased exposure to sunlight and the fact that non-fortified foods do not contain enough VD. Food fortification is a practical solution for addressing the problem of VD deficiency. Here we discuss the most recent findings about the relationship of VD deficiency and the cardiovascular system as well as the latest advances in food fortification with VD. This work has been funded by the program "Cooperation", Measure 16, of the Hellenic Ministry of Rural Development and Food.

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Multi-omics technologies and AI models applied to animal science

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Next-generation sequencing (NGS) technologies have substantially

improved the methods of omics analyses (genomics, transcriptomics, metabolomics, metagenomics). These methods are powerful and valuable tools in various fields such as the study of genetic diseases, metagenomics and animal breeding. Nowadays, multi-omics methods are gaining popularity in animal science due to their ability, compared to single analyses, to extract and interpret complex and hidden information. Multi-omics analyses can be analysed by creating a network between different omics data. The network serves as a mathematical model to describe the complex interactions and regulatory mechanisms that occur in biological systems. Multi-omics data can also be analysed with predictive algorithms based on Machine Learning (ML) or Deep Learning (DL). ML and DL approaches allow for a high degree of processing and modelling capacity of omics data. These models can effectively solve certain problems such as class imbalance, high dimensionality, data scalability and missing data. Before analysing multi-omics data, it is, however, necessary for each omics data to carry out data imputation and outlier detection separately. Furthermore, despite the progress made in various disciplines of animal sciences in multi-omics analysis and AI, many challenges still remain. These include the analysis of large amounts of multi-omics data.

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Pharming Animals: Exploring the Advantages and Disadvantages of Biopharmaceutical Production through Transgenic Livestock

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Transgenic farm animals are becoming integral to human medicine, serving as valuable sources of biologically active proteins, participants in xenotransplantation, and subjects for extensive research in cell and gene therapy. In the agricultural sphere, these animals offer opportunities to improve carcass composition, boost lactational performance, enhance wool production, fortify disease resistance, and mitigate environmental impact. To ensure the safety and quality of transgenic animal products, standardization of procedures is paramount, reinforced by polymerase chain reaction and array technology. The advent of advanced techniques, including somatic nuclear transfer, has supplanted conventional methods like DNA pronuclear microinjection, allowing for greater efficiency and precision in transgenesis. However, the path to achieve the full potential of pharming animals is fraught with ethical dilemmas, intricate regulatory frameworks, public perception and acceptance hurdles, potential environmental risks, and substantial initial investments required to establish transgenic animal facilities. These challenges demand a comprehensive and collaborative approach that emphasizes transparent communication, research and development efforts, economic incentives, and consumer education. In this regard, this study aimed to realise a thorough investigation into the pros and cons of utilizing pharming animals for biopharmaceutical production, with the objective of imparting a nuanced comprehension of transgenic livestock's role in this sector.

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Proposal of a mathematical model based on the integration of multi-omic and clinical data to predict individual responses to nutrition and exercise programs

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Optimizing athletic performance requires precise personalization of diet and exercise. This study aims to develop an innovative mathematical model that integrates multi-omics (genomics, proteomics, metabolomics) and clinical data to predict individual responses to nutrition and training programs, optimizing body composition in terms of lean/fat mass ratio. Data were collected from 515 athletes, including parameters of body composition, physical activity, and caloric intake. In anticipation of the multiomics analyses, and by studying the models already in the literature and the nature of the data to be modeled, the mathematical model for their analysis was developed. Unlike classical models that use only clinical data or a single omics data type, our approach uses advanced supervised machine learning techniques, including regression models and neural networks, to integrate multi- omics and clinical data. The model pipeline includes normalization and data imputation, clustering to identify homogeneous groups, and prediction of individual responses. To date, few models have been built to integrate multi-omics and clinical data in this area, and therefore this study is a first attempt to compare and improve existing models.

Keywords: Mathematical models, Omics data, Clinical data, Machine learning

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Polygenic risk score study of a cohort of 135 anorexic patients proposes new genetic polymorphisms that expand the architecture of the genetic basis of the disease

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The Polygenic Risk Score (PRS) estimates the common genetic component of an individual towards a specific phenotype. This score considers the cumulative effect of various SNPs, providing an assessment of individual risk to develop a disease. Our study, based on published algorithms, calculated the PRS to understand the role of SNPs in AN. Targeted sequencing of 730 SNPs was performed on 135 anorexic patients and 98 controls, followed by the construction of a machine learning

model to predict PRS using logistic regression. We evaluated the Hardy-Weinberg Equilibrium and, using logistic regression with Plink and PRSice, we validated the selected SNPs in two independent case-control studies for the association to AN. Results showed a significant difference in PRS scores between patients and controls, -0.15 and -0.32, respectively. The PRS had an R-squared value of 0.213 and a p-value of 2.1e-57, demonstrating an association between the calculated PRS and AN risk. From this study, new genetic polymorphisms in the genes MCM6 and CNDP1 have emerged, allowing the expansion of the genetic architecture of anorexia.

Keywords: Polygenic Risk Score, Anorexia nervosa, SNPs, Personalized risk assessment

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Mathematical model for oncological screening using mass spectrometry data from the analysis of tumor specific peptides in serum

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Cancer screening employs specific techniques such as imaging, biochemical and omics analysis. This study explores the integration of mass spectrometry with machine learning algorithms to classify and detect various cancer types early by analyzing tumor-specific peptides in serum. A curated dataset composed of 200 patients with four main types of cancer (lung, breast, prostate and colon) was considered. Standardization was applied for feature scaling, optimizing model performance. The study employed Support Vector Machine (SVM) with a Radial Basis Function kernel, Random Forest (RF), and Logistic Regression (LR) for supervised classification. Serum samples underwent elaboration to isolate peptides, which were analyzed using high-resolution mass spectrometry. The resulting peptide profiles allowed for precise differentiation between cancerous and non-cancerous samples. The RF model achieved the highest classification accuracy at 95%, followed by SVM at 92%, and LR at 89%. The main identified tumor-specific peptides comprised PAP, PSA, HER-2/neu, Kallikrein 4, and mdm-2. Although this study needs to be validated on a larger cohort of patients, it underscores the potential of mass spectrometry combined with machine learning for non-invasive cancer screening.

Keywords: Mass spectrometry, Machine learning, Tumor-specific peptides, Non-invasive cancer screening

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Mathematical model based on mutational loading to estimate the genetic component in Inherited Retinal Dystrophies patients negative for Mendelian genetic testing

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Inherited retinal dystrophies (IRDs) are diseases characterized by progressive vision loss with partially known genetic causes. However, traditional genetic testing based on Mendelian inheritance fails to diagnose 25-40% of cases. This study hypothesizes that the undiagnosed cases also involve interactions between allelic variants across multiple genes, which are not considered by conventional models. A mathematical model was developed to assess mutational load in patients with negative genetic testing for IRDs. Were analysed 453 patients and 74 controls, focusing on genetic variants in metabolic pathways. Variants were weighted according to ACMG class (VUS=0.2, LP=0.3, P=0.5) and mapped to relevant pathways. Statistical analyses, including Mann-Whitney U tests and Kernel Density estimates, identified significant pathway activations, "Retinoid Cycle Disease Events" pathway was strongly associated. Considering the 10% of the case with the highest score as positive, the cut-off score ≥1.32 was established for sample positivity. The results indicate that advanced computational models can help discover new genetic interactions responsible for IRDs considering oligogenic inheritance. This theoretical model will need to be validated with segregation and functional studies.

Keywords: Mutational loading, Retinal dystrophy, Genetics.

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Mathematical model based on plasma proteomic profiles for active and latent phase classification of patients with hereditary retinal dystrophies

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Hereditary retinal dystrophies include conditions such as inherited maculopathies, retinitis pigmentosa, cone and rod dystrophies, and age-related macular degeneration, all of which are characterized by progressive visual loss. The correlation between disease progression and clinical diagnosis is often unclear, and variations in retinal degeneration and visual loss can differ greatly among individuals with the same genetic mutation. This study aims to examine by mass spectrometry plasma proteins related to retinal function in healthy subjects to establish a baseline. We analyzed 324 proteins in the plasma of 500 healthy individuals, divided by gender and age groups between 40 and 60 years. The proteins studied included retinal structural proteins, retinoid cycle proteins, mitochondrial proteins, proteins involved in apoptosis, inflammation, immunity, oxidative stress, and plasma transport. The analysis revealed variations in protein levels with age and differences between genders, identifying a reference proteomic map for different clusters. By comparing these data with those of subjects with retinal dystrophies, the study aims to identify the stages of the disease into latent and active, to allow better prediction of disease evolution.

Keywords: Machine learning, proteomics, retinal disease.

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Integrative multilevel genetic evaluation in cardiovascular disease: Mendelian genetics, Polygenic Risk Score and pharmacogenetics approaches

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Cardiovascular diseases are the leading cause of death in adults and represent a significant global health challenge. Traditional Mendelian diagnostic tests explain only the 10-20% of the genetic basis of cardiomyopathies and arrhythmias. Using next-generation sequencing, this study investigates in 312 patients diagnosed with cardiovascular diseases a comprehensive genetic panel to concurrently assess Mendelian traits (170 genes), polygenic risk scores (977 SNPs), and pharmacogenetics traits. The Mendelian genetics panel covers cardiomyopathies, including arrhythmogenic, dilated, restrictive, hypertrophic types, genetic defects leading to malformations of the heart and heart valves, and Mendelian vascular disorders. The PRS panel assesses the multifactorial aspect, identifying the risk for myocardial infarction, sudden death, and post-COVID-19 complications. The pharmacogenetics panel includes genes associated with hyperlipidemia, hyperhomocysteinemia, cardiotoxicity, and other heart-related conditions, enabling the identification of tailored anti-platelet, cholesterol-lowering therapies, and identification of individuals at risk for chemotherapy-induced cardiotoxicity. By integrating data from these panels, we can provide a comprehensive genetic landscape of cardiovascular patients, enhancing our understanding of cardiovascular disease genetics and improving patient management.

Keywords: Cardiovascular diseases, Genomics, PRS, pharmacogenetics

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Predictive mathematical model for identification of lipedema in subcutaneous adipose tissue biopsies through integration of clinical and multi-omics data

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Lipedema is a chronic condition characterized by abnormal accumulation of subcutaneous adipose tissue, which causes pain, swelling, and psychological distress. Differentiating lipedema from other disorders that are associated with subcutaneous adipose tissue accumulation, such as obesity, lymphedema, lipodystrophies, and Dercum's disease, is difficult because of overlapping symptoms and multifactorial etiology. This study aims to develop a mathematical model using advanced supervised machine learning techniques to analyze multi-omics data, including metabolomics and proteomics, to identify specific biomarkers and molecular pathways indicative of lipedema, thereby improving diagnostic accuracy. We employed a novel approach by integrating demographic, clinical and multi-omics data from 63 patients with lipedema, comparing affected and healthy adipose tissues. Differentially expressed key metabolites and proteins, such as histamine and ACSL1, were identified. The mathematical model incorporates feature selection algorithms and dimensionality reduction methods. Our model demonstrates the potential of integrating multi-omics data with mathematical approaches to refine the differential diagnosis of lipedema, offering a contribution in the identification and understanding of this complex disease.

Keywords: Lipedema, Machine learning, Proteomics, Metabolomics

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Cornering stroke diagnostics with electrolateral flow quantitation (POC4triage)

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Stroke (part of the “First Hour Quintet”), a leading cause of death in the EU, necessitates the ambulance to take the patient to the appropriate care service due his life-threatening state, where every minute is critical to improve his outcome. In addition, the ambulatory prehospital identification ought to be relayed to the hospital emergency medical services before arrival of the patient to ensure crucial lifesaving clinical decision making. Consequently, we embarked in developing a portable stroke-specific kit to detect its relevant blood biomarkers based on our proprietary electrolateral flow immunosensor that is designed to be specific, provide accurate quantification, enables multiplexing (several biomarkers), is compact, easy to use, robust, and cost-effective, and from which patient data is sent rapidly to the receiving hospital information system, thus, entering the clinical workflow, to help triage patients upon arrival to the appropriate care unit soonest and ensuring readiness to receive the patient. The technology edge is its inherent quantitation ability, eliminating a major drawback in POCTs, without compromising in sensitivity (microelectrode electrochemistry), while allowing one step convenience test (lateral flow flow technology). This hybrid technology was made possible by our electroactive immuno- specific nanoparticles and their quantitative reading made by a hand-held galvanometer from a drop of blood. Our proof-of- concept prototype (second generation) was made possible using a model biomarker, NT-proBNP for which we obtained a limit of detection (LOD) of 250 pg/ml.

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Artificial intelligence in clinical genomics: Phenotype-to-genotype

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The shift from analog to digital technologies in clinical laboratory genomics is bringing about a "big data" era, surpassing human capacity to efficiently analyze data with traditional methods. To accurately interpret complex molecular data for diagnosing and managing genomic disorders, artificial intelligence (AI) will play a crucial role. AI is already being used in genomics to detect DNA variants, predict how these variants affect protein structure and function to assess pathogenicity, connect phenotypes with genetic variants from exome or genome sequencing for quicker diagnoses, align genomic data with cancer staging and treatment plans, use natural language processing (NLP) to quickly access key medical literature, and deploy chatbots to assist with genetic testing eligibility and pre- or post-test education. With ethical and thorough development, AI is set to enhance the ability of geneticists to distill complex data into clear insights for clinicians, enabling more effective patient care on a larger scale. In my talk, I will talk about the artificial intelligence models we have developed for the diagnosis of complex genetic disease groups such as cancer and ciliopathies.

Keywords: artificial intelligence, genotype, phenotype, genetic disease, genomics