

**ESRT**  
**4TH INTERNATIONAL**  
**MEDICAL STUDENTS'**  
**CONGRESS 2020:**  
**GENETICS**

**ABSTRACT BOOK**



## **Congress Information**

**Date:** 14-16 March 2020

**Location:** Ege University Faculty of Medicine, Muhiddin Erel  
Congress Hall

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## FOREWORD

Ege Scientific Research Team (ESRT) was established in 1987 by a group of Ege University Medical Faculty students in order to learn scientific thinking and scientific research methods with a multidisciplinary approach.

Students who came together to do and learn scientific research under the roof of ESRT conducted many studies in 30 years. Until today as a result of the studies that ESRT there have been above 15 international and 50 national publishes. As ESRT, our sole purpose is not to learn knowledge but also to share that. For this purpose, we have organized many national and international activities up until today. If we look at these activities there have been 3 international, 5 national congresses and 7 summer/winter camps that aim to share knowledge. This year we are happy to be with you in our 4th International Medical Students' Congress 2020: Genetics.

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*We wish that our programme will go well for our precious participants.*

Ege Scientific Research Team

## **RECEPTION AND ACKNOWLEDGEMENTS**

### **Reception**

In our abstract book, there are oral and poster presentations' abstracts for our precious participants.

### **Acknowledgements**

We appreciate our Supervisor Faculty Members for being here all the time and supporting us by sharing their experiences.

Also we appreciate Ege University Rectorate, Ege University Faculty of Medicine Deanery and Directorate of Health, Culture and Sports for letting us use the sources of our university and helping us at any need.

### **Sponsorships**

We thank Izmir Metropolitan Municipality, Cafe Tıp, Akça Medikal and Alp Erdem Elektrik for their support.

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# SCIENTIFIC PROGRAMME

Saturday, 14 March 2020

Registration: 9.15-9.45

Opening Ceremony: 9.45-10.15

1st Session: EPIGENETICS

10.30-11.00 Guest Speaker: Prof. Dr. Stefan DIMITROV: Epigenetics, Life Beyond DNA

11.00-11.15 Coffee Break

11.15-11.35 Guest Speaker: Prof. Dr. Ali HAMICHE: Chromatin and Epigenetic Regulation in Cancer

11.35-12.30 EVALUATION OF POSTER PRESENTATIONS

Lunch Break: 12.30-13.30

2nd Session: MOLECULAR GENETICS

13.30-14.00 Guest Speaker: Prof. Dr. Jacob HANNA: Human-Mouse Cross-Species Chimerism

14.00-14.15 Oral Session: Enes Salih ARPA: Importance of NAD<sup>+</sup> and Boosting Strategies via Targeting NAD<sup>+</sup> Pathway Genes

14.15-14.25 Coffee Break

14.25-15.05 Guest Speaker: Prof. Dr. Halil KAVAKLI: Gene Expression and Circadian Rhythm

15.05-15.20 Coffee Break

15.20-16.00 Guest Speaker: Prof. Dr. David BURSTEIN: Treasures in the Wild, Novel CRISPR-Cas from Uncultivated Microbes

16.00-16.15 Oral Session: Belkis Aysu ÖZBEK: Control of Genome Editing Cas9 Protein with Oxygen

16.15-17.30 WORKSHOPS



Sunday, 15 March 2020

1st Session: CLINICAL & PEDIATRIC GENETICS

9.30-10.10 Guest Speaker: Prof. Dr. Lucy RAYMOND: The Genetic Causes of Intellectual Disability

10.10-10.25 Oral Session: Laman HUSEYNLI, Javid MUSTAFAZADA: Rare Case of the Clinical Combination of Two Nosological Forms of Chromosomal Pathology

10.25-10.40 Coffee Break

10.40-11.10 Guest Speaker: Prof. Dr. Ahmet Okay ÇAĞLAYAN: Applications of New Generation Sequencing Technologies in Medical Genetics Research

11.10-11.25 Oral Session: Kardelen GÜLEÇ: Genetic Role of Nutrition in Diseases

11.25-11.40 Coffee Break

11.40-12.10 Guest Speaker: Dr. Anıl BİRİCİK: Preimplantation Genetic Testing (PGT): Current Applications and Future Perspectives

Lunch Break: 12.10-13.30

2nd Session: ONCOGENETICS

13.30-13.55 Guest Speaker: Dr. İbrahim Gökçe YAYLA: Introduction to Electrochemotherapy: Principals, Mechanism of Action, Application to Cancer Treatment and The Turkish Experience

13.55-14.10 Oral Session: Merve EROĞLU, Çiğdem EROĞLU: Case Presentation of NF1

14.10-14.25 Coffee Break

14.25-15.05 Guest Speaker: Prof. Dr. Emin KANSU: Tumor Evolution in the Genome Age and Darwin

15.05-15.20 Oral Session: Ceren YÜRÜMEZ: Alu Elements and Their Role In Population Genetics

15.20-15.55 Award & Closing Ceremony

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# **ORAL PRESENTATIONS**

## **Epigenetics, Life Beyond DNA**

*Prof. Dr. Stefan DIMITROV*

Epigenetics is the study of cellular and physiological trait variations that are not caused by changes in the DNA sequence (a change in phenotype without a change in genotype).

Epigenetic changes occur naturally but can also be influenced by several factors including age and the environment/life style. Strong alterations in epigenetic features are associated with numerous diseases, including cancer. In contrast to genetic alterations, the epigenetic alterations are, in principle reversible, and could be targeted by specifically designed drugs.

My talk will be concentrated on chromatin epigenetics. It will summarize literature reports and data from our lab. After an introduction on the main aspects of structural epigenetics, I will focus on the epigenetic mechanisms and strategies that the cell is using to control vital processes. How these mechanisms are then altered in diseases and in particular in cancer, will be next described and discussed. Finally, I will show examples of specific depletion of epigenetic factors in various organs in conditional knock out mouse models and the resulting phenotypes. These experimental data will reveal in vivo the roles of these factors in preserving cell homeostasis.

## **Chromatin and Epigenetic Regulation in Cancer**

*Prof. Dr. Ali HAMICHE*

Rett syndrome is an X-linked autism-spectrum disorder caused by mutations in MECP2, encoding methyl CpG-binding protein 2. Since the discovery of MECP2 mutations as the genetic cause of Rett syndrome, the understanding of MeCP2 function has remained elusive. Twenty-eight years of research have painted a complex picture of MeCP2 function. However, despite of the thousands studies and the enormous efforts invested by both researchers and clinicians, the MeCP2 roles in normal brain and in Rett syndrome remains far from being clear. During this presentation I will clarify some fundamental features of MeCP2 functions under both normal and pathological conditions and I will discuss how epigenetic regulation, chromatin organization and circuit dynamics contribute to the onset of Rett syndrome.

## **Human-Mouse Cross-Species Chimerism**

*Prof. Dr. Jacob HANNA*

The identity of somatic and pluripotent cells can be epigenetically reprogrammed and forced to adapt a new functional cell state by different methods and distinct combinations of exogenous factors. The aspiration to utilize such ex vivo reprogrammed pluripotent and somatic cells for therapeutic purposes necessitates understanding of the mechanisms of reprogramming and elucidating the extent of equivalence of the in vitro derived cells to their in vivo counterparts.

In my presentation, I will present my group's recent advances toward understanding these fundamental questions and further detail our ongoing efforts to generate developmentally unrestricted human naive pluripotent cells. I will conclude by highlighting new avenues for utilizing epigenetic reprogramming to naïve pluripotency for unraveling critical gene regulatory mechanisms acting during early mammalian development and highlighting prospects for new platforms for human disease and developmental modelling.



## **Importance of NAD<sup>+</sup> and Boosting Strategies via Targeting NAD<sup>+</sup> Pathway Genes**

Enes Salih ARPA

**Background:** Nicotinamide adenine dinucleotide (NAD<sup>+</sup>) is a cosubstrate for several enzymes, including the sirtuin family of NAD<sup>+</sup>-dependent protein deacylases. Beneficial effects of increased NAD<sup>+</sup> levels and sirtuin activation on mitochondrial homeostasis, organismal metabolism and lifespan have been established across species. Here we show that, expression of NAD<sup>+</sup> pathway genes are different for each organ.

**Aim:** Targeting of that genes may ameliorate some organ specific diseases (e.g. NAFLD for liver).

**Methods:** Several organs were used to see NAD<sup>+</sup> pathway gene mRNA expression difference across tissues for DBA/2J mice.

These organs were; liver, left ventricle of heart, subcutaneous white adipose tissue, spleen, kidney, gastrocnemius and quadriceps. RNA extraction was performed on all of these tissues. Following that, cDNA synthesis and quantitative real time PCR were performed for all samples.

**Results:** For the most of genes; liver was showed the best mRNA expression. For only ACMSD gene; kidney was showed the best mRNA expression. For sirtuins; muscle tissues were showed good expression. ( $p < 0.05$ ).

**Conclusion:** The mRNA expression of NAD<sup>+</sup> pathway genes are different for each organ. If we purpose to ameliorate NAD<sup>+</sup> based diseases, we may target specific genes for each organ (e.g. ACMSD for kidney diseases).

**Keywords:** NAD<sup>+</sup>, mRNA-expression, DBA/2J mice, ACMSD, qPCR

**References:**

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## Gene Expression and Circadian Rhythm

Prof. Dr. Halil KAVAKLI

Many biochemical, physiological and behavioral processes in living organisms show oscillations in periods of ~ 24 hours. Circadian rhythms include activity/rest, nutrition /hunger, reproduction, body temperature, certain hormones and metabolite cycles. The Mouse studies showed that 43% of the genes encoding protein in all tissues are under control of circadian rhythm. At the molecular level, the circadian rhythm main clock proteins CLOCK interacts with the BMAL1 protein to form a dimer. The CLOCK-BMAL1 pair binds to the E-box located in the promoter region of the genes and initiates the expression of the genes in the clock control to form the positive cycle of the mechanism. Cryptochrome (CRY) and PERIOD (PER) proteins, which begin to accumulate in the cytoplasm, pass into the nucleus to form a complex with CLOCK-BMAL1 and form a negative cycle of the mechanism by repressing the genes under the control of the clock. Both cycles provide a 24-hour rhythm in the cells. I will talk about how circadian clock regulates so many different physiological variables at molecular level.

## **Treasures in the Wild: Novel CRISPR-Cas Systems from Uncultivated Microbes**

*Dr. David BURSTEIN*

Current understanding of microorganism–virus interactions, which shape the evolution and functioning of Earth’s ecosystems, is based primarily on cultivated organisms. We applied a cultivation-independent approach to seek new CRISPR-Cas systems that will contribute to our understanding of the arms-race between viruses and their microbial hosts, and will potentially expand the genome editing toolbox. Using genome-resolved metagenomics, we identify a number of novel CRISPR–Cas systems, these include streamlined types of CRISPR-Cas systems found exclusively in uncultivated microbes, tiny systems targeting single-strand DNA, and the first reported Cas9 in the archaeal domain of life. Notably, all required functional components were identified by environmental metagenomics and metatranscriptomics, enabling validation of robust in vivo RNA-guided DNA interference activity in *Escherichia coli*.

## **Control of Genome Editing Cas9 Protein with Oxygen**

*Belkıs Aysu ÖZBEK*

**Background:** Mutations can cause beneficial species differences, but it can also result in many genetic disorders. CRISPR is new, efficient, easy and cheap method that is targeted to genome editing. There is a need for a Cas9 protein which is not active all the time in the cell because it can cause serious problems by targeting genome regions.

**Aim:** I aimed to produce a Cas9 protein with recombinant DNA technology.

**Materials:** VP12 plasmid, DNA isolated from blood, eGFP plasmid, BamH1 Xho1 Hind3 restriction enzymes, waterbath incubator, PCR product.

**Method:** ODD domain synthesis increase in the hypoxia condition, using this feature I designed a plasmid that has ODD domain and can produce Cas9 protein in the absence of oxygen and then added eGFP gene into plasmid. The role of eGFP protein in here is to determine if plasmid has reached to the nucleus of the cell using its feature of fluorescent under microscope. After transfecting Cas9-eGFP-ODD protein into the cell line they incubated in the hypoxia conditions.

**Results:** I designed and generated plasmid capable of forming a modified Cas9 protein which can be synthesized in oxygen poor cells. I have created a plasmid that could form the Cas9-eGFP-ODD protein.

**Conclusion:** This designed biotechnological tool and method provides control of Cas9 activity with harmless and natural oxygen. This in vitro conditions, this tool may allow genome correction in cell lines and patient cells to be more controlled and easier.

**Keywords:** recombinant DNA, CRISPR/CAS9, HIF, hypoxia, ODD, GFP, plasmid

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## **Genetic Causes of Intellectual Disability**

*Prof. Dr. Lucy RAYMOND*

The underlying etiology of Developmental Delay or Intellectual Disability has changed dramatically since the advances in vaccination, obstetric and perinatal care. However, as neonatal care and management of the premature infant has improved these are increasingly recognised to contribute to intellectual development. Nevertheless, the genetic contribution to intellectual disability is recognised as the predominant cause. The rate of diagnosis of children with intellectual disability has improved as the ability to perform DNA sequence analysis at scale. Mechanisms of disease and methods of detecting mutations will be discussed and illustrated throughout with genetic forms of ID. The use of gene agnostic trio analysis of genomes will be presented as a comprehensive method of identifying most known causes of disease. The challenge remains to identify the remaining genes that cause disease and to develop strategies of therapeutics to cope with the thousands of genes that have been characterised to date.

## **Rare Case of the Clinical Combination of Two Nosological Forms of Chromosomal Pathology**

Laman HUSEYNLI, Javid MUSTAFAZADA

**Background:** Chromosomal pathology makes a significant contribution to perinatal morbidity and mortality. Only few options of numerical chromosome abnormalities in children are compatible with postnatal development and lead to chromosomal diseases. Aneuploidy is the second most important category of chromosome mutations relating to abnormal chromosome number.

**Aim:** The research presents a rare case of the clinical combination of two nosological forms of chromosomal pathology in a patient.

**Materials and methods:** Clinical description of a case of combined chromosomal pathology: Down syndrome and trisomy on the X chromosome in patient A. The cytogenetic conclusion obtained by standard karyotyping with differential staining of chromosomes is 48, XXX, +21, which corresponds to the presence of two additional chromosomes the X chromosome and the autosome 21.

**Results:** The proband simultaneously diagnoses two nosological forms of chromosomal pathology: Down syndrome and trisomy on the X chromosome

**Conclusions:** The case of combined chromosomal pathology in a child A. indicates the importance of monitoring of the fetus conditions in the first and second trimesters of pregnancy for the timely detection of fetal malformations and, if necessary, invasive prenatal diagnosis of chromosomal pathology in a specified period to the further decision on prolongation or interruption of a pregnancy with an abnormal fetus.

**Keywords:** down syndrome, trisomy X, malformations, chromosomal syndromes



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# **Applications of New Generation Sequencing Technologies in Medical Genetic Research**

*Prof. Dr. Ahmet Okan AĞLAYAN*

Neurodevelopmental disorders (NDDs) are a heterogeneous group of conditions that carry significant mental, emotional, physical, and economic consequences for individuals, their families, and society in general. The substantial genetic and phenotypic heterogeneity of neurodevelopmental disorders has made the identification of candidate genes very challenging. Next-generation sequencing is transforming the field of clinical genetics providing a new opportunity for gene discovery in NDDs. Identifying the disease causing genes will advance understanding of the molecular mechanisms underlying these disorders and provide insight into potential therapeutic interventions. In the past 10 years, I have involved and led a series of successful studies identifying novel disease causing genes in structural brain diseases using next generation sequencing techniques. I was a co-author several papers published in Brain, Cell, Nature, Science, Nature Genetics, Neuron and PNAS, reporting new disease genes in NDDs, identified through next generation sequencing. I'll share these studies with the audience.

## **Genetic Role of Nutrition in Diseases**

Kardelen GÜLEÇ

Due to changing in daily diets in modern life probably increase the related diseases in human mankind. This situation has pushed scientists to seek new ways to prevent these diseases. Genetic structure, which is the first factor in the pathogenesis of each disease, is located at the beginning of these searches. Environmental factors have been proven to play a role in the regulation of DNA methylation, histone modification, and gene expression via non-encoded RNA, although there is no direct structural effect on genetic material. Therefore, epigenetics is gaining importance with the increasing number of studies in recent years.

The completion of the Human Genome Project in 2003 has been an important milestone in nutrition and genetics, as in some other sciences. As a result of the developments in the field of nutrition and metabolism, it has been understood that nutrient components directly or indirectly affect gene expression, thus metabolism and health. Scientists started questioning if the interaction between genes and food bioactive compounds could positively or negatively influence an individual's health. In order to assess this interaction between genes and nutrients, the term “Nutrigenomics” was created.

In our presentation; we aim to evaluate the relationship between genetics and nutrition, nutrigenomics field which has just entered the literature through various studies and living samples.

Keywords: epigenetics, nutrigenomics, diseases,

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## **Preimplantation Genetic Testing (PGT): Current Applications and Future Perspectives**

*Dr. Anıl BİRİCİK*

Preimplantation genetic testing (PGT) is a procedure used prior to implantation of IVF (In Vitro Fertilization) embryos and provides a reproductive option to the couples at genetic risk to avoid having an affected pregnancy. Currently PGT is being applied in clinical IVF for two aims: 1) To reduce the genetic risk, in patients suffering from a genetic disorder 2) To increase clinical IVF outcome by selecting euploid embryos. The first step of PGT is embryo biopsy and this can be done on different developmental stages of IVF embryos starting from pre and post-fertilization stages by polar body biopsy, at cleavage stage by blastomere biopsy and at blastocyst stage by trophectoderm (TE) biopsy followed by whole genome amplification reaction (WGA) for downstream applications. Together with current standard PGT protocols like single cell multiplex PCR and array comparative genomic hybridization (aCGH), a new technology next generation sequencing (NGS) has successfully adapted in PGT field with its potential to deliver high throughput genetic data in a short period at lower cost. The flexibility of the NGS technology allows us to design experimental protocols according to our needs.

For instance, combined applications can be done on multiple samples in the same NGS reaction. The high sensibility of such brand new technologies open new application fields to test alternative DNA sources for PGT such as cell-free embryonic DNA in spent culture media or embryonal blastocoel fluid. Even if the clinical use of these approaches is still early, the initial data emerged from the first studies are promising. However, it should always be taken into consideration that each new application can be introduced in clinical practice only after well-designed validation studies, and corresponding technical and ethical guidelines should always be followed to provide the correct counselling and the treatment to the patients.

## **Introduction to Electrochemotherapy: Principals, Mechanism of Action, Application to Cancer Treatment and The Turkish Experience**

*Dr. İbrahim Gökçe YAYLA*

In the first part of this presentation, the principle of electrochemotherapy (ECT) will be presented including its mechanism-of-action, its equipment as well as its application to the treatment of multiple malignancies. In the second part of the presentation, a review of world-wide results using ECT will be presented including the results of clinical trials and case studies. In the third part, a series of ECT applications to Turkish patients will be presented with before and after images. Finally, in the last part, the future of ECT will be discussed including gene therapy and application to deep-seated tumors.

## Case Presentation of NF1

Merve EROĞLU, Çiğdem EROĞLU

Neurofibromatosis 1 (NF1) is characterized by multiple café au lait spots, axillary and inguinal freckling, multiple cutaneous neurofibromas, iris Lisch nodules, and choroidal freckling. About half of people with NF1 have plexiform neurofibromas, but most are internal and not suspected clinically. Learning disabilities are present in at least 50% of individuals with NF1. Less common but potentially more serious manifestations include optic nerve and other central nervous system gliomas, malignant peripheral nerve sheath tumors, scoliosis, tibial dysplasia, and vasculopathy. Approximately one-half of the cases are familial(inherited) .Our cases have a novel mutation on NF1 gene c.4991\_4997del TTGTTTT and include most of the symptoms of NF1.

Our findings indicate that NF1 is an autosomal dominant disease and we experience this disease quite frequently. Therefore our aim is try to make audiences be awareness of NF1 and its importance.In this case patients who have NF1 might get a diagnosis earlier thus providing reduce of malpractice and the treatments can possibly much more successful.

Keywords: genetics, NF1, disease, patient

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## **Tumor Evolution in the Genome Age**

*Prof. Dr. Emin KANSU*

## **Alu Elements and Their Role in Population Genetics**

Ceren YÜRÜMEZ

Alu elements are sequences of DNA which are making up approximately 10% of the human genome. They can be classified as SINE's which are non-autonomous retrotransposons. Also Alu elements are primate-specific transposable elements. They insert copies of themselves into protein-coding genes

Alu elements transcribe into mRNA and then converted into a double stranded DNA by reverse transcriptase. The new DNA is inserted into a different location in genome.

The probability of two different Alu elements inserting into same place independently is zero and newly inserted Alu elements undergo deletion rarely. These make Alu insertion polymorphism perfect for genetic markers to use in population genetics.

By analysing Alu insertion frequencies among different populations, we can learn how humans spread through the world. Alu diversity is highest in Africans, lowest in Europeans and Alu insertion frequency is lowest in Africans, highest in Indians, East Asians and Europeans. Large genetic distances are observed among African populations and African and non-African populations. The root of a neighbor-joining network is located closest to African populations. These findings show that human populations have an African origin and human populations left Africa to colonize the world.

Keywords: Alu elements, retrotransposons, population genetics, Alu insertion

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# **POSTER PRESENTATIONS**

# **The Future of Genetic Science: Analyses of Genomal Regulation and Development in CRISPR Technology**

*Merziye Gökçe GÖKSU, Müge KARAKAYALI*

We wanted to show the development of CRISPR technology, which is the future of genetics in our project, and to introduce this technology early. CRISPR/Cas9 is a technology that can change the genetic code. We examined the development of this technology from "Nature" magazine. We have found that CRISPR technology has great potential from the moment it was found and scientists have been trying to impose some limitations on the use of this technology from the very beginning.

However, in China, the controversy has gained momentum with the scientist named He Jiankui changing the genome of twin babies and making them HIV resistant. As a result, scientists organized international panels and introduced various laws in different countries. He was fired from his job. And some firm decisions were made by scientists. Since we do not know the long-term effects of changing a gene, international laboratories are not currently allowed to transition to human experiments, but animal experiments are ongoing. This technology, which is very useful in terms of medicine, will come across frequently in the coming years and will bring both scientific revolutions and ethical problems.

Keywords: genetics, CRISPR, genetic code, ethic

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# **The Importance of NER Mechanism on DNA Stability and Human Syndromes**

Didar FATTAHI, Sahra ESMKHANI

This article investigates the DNA repair mechanisms and its relation to certain diseases. Nucleotide excision repair (NER) of DNA is responsible for genome maintenance and the overall organism preservation. Studying cells from recessive genetic disorders has led to better understanding of the NER mechanism. Some of these rare genetic disorders include: xeroderma pigmentosum, trichothiodystrophy, and Cockayne syndromes. Cells from these disease states have some commonality, which is impaired capacity of repairing UV-induced DNA lesions. This article mainly focuses on essentials of NER and how impairment in capacity and function lead to related human genetic disorders.

**Keywords:** DNA damage response, nucleotide excision repair, transcription-coupled repair, XP, CS, TTD

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# Co-occurrence of 6q25 Microdeletion and 14q32 Microduplication in a Dysmorphic Child

*Hazal UZUN, Seyma ACAROĞLU*

**Introduction and purpose:** Balanced translocations are the most common structural chromosomal abnormalities in humans. Unbalanced products in the gametogenesis of balanced translocations may cause microdeletions and microduplications in the next generation. The 6q25 microdeletion and 14q32 microduplication are distinct entities with distinct clinical features. In this study, we will discuss the co-occurrence of these two genetic anomalies in a family of balanced translocation carriers, which have not been previously reported in the literature.

**Case:** A 4-year-old girl was referred to us because of growth and developmental delay and dysmorphic appearance. Ventriculomegaly was detected on antenatal ultrasonography (USG) and she was operated postnatally due to congenital anal stenosis. In the physical examination of the patient, microcephaly, bitemporal narrowing, scaphocephaly, wide forehead, high anterior hairline, hypertelorism, sparse hair, eyebrows and eyelashes, periorbital edema, thin upper lip, micrognathia, flattened nasal root, anteverted nares, bulbous nasal type, prominent premaxilla, wide mouth and downward corners of mouth, hypotonia, cliteromegaly were noted. Cranial MR revealed corpus callosum agenesis and colpocephaly. Abdominal USG showed adrenal gland enlargement and adrenal MRI was consistent with bilateral adrenal gland hyperplasia. Echocardiography showed an atrial septal defect. Liver position, size and echogenicity; gallbladder wall and lumen were normal.

**Results:** Array-Comparative Genomic Hybridization (aCGH) is performed from peripheric blood and a microdeletion of 10.88 Mbp between 6q25 and 6q27 involving 41 OMIM gene and a microduplication of 11.06 Mbp containing 73 OMIM gene on chromosome 14q32 were detected.

**Discussion:** In recent years, the effect of Array-CGH technology has enabled the identification of many new syndromes in patients presenting with intellectual disability, dysmorphic findings and multiple congenital anomalies. We believe, the co-occurrence of 6q25 microdeletion and 14q32 microduplication in a patient with distinct congenital and dysmorphic abnormalities, is a new entity introduced to the literature by this publication.



Keywords: microduplication, microdeletion, unbalanced product

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# **What Should The Ethical Rules Be After CRISPR Edited Babies Born**

Zehra İLİMAN, Salar GACHPAZ SARKHIZ

**Background:** Clustered regular interval short palindromic repeats (CRISPR)-(Cas) 9 has been used to collect functional areas that suppress/activate gene expression in living cells or organisms or label specific genomic loci in living cells or organisms to investigate gene expression regulation and effects on the human embryo. Studies on the human embryo may produce different mutations that have unknown effects. Furthermore, embryo editing is only ethically justifiable in cases where the benefits clearly outweigh the risks. Is it necessary to reorganize the ethical concepts discussed on babies edited by CRISPR.

**Aim:** The aim of this study is to investigate the ethics of gene editing and results by CRISPR on the human embryo.

**Methods:** It is processed that discussed issues about embryo gene-editing after new developments.

**Results:** Several media outlets report that a team of scientists in China has used CRISPR to modify the DNA of healthy human embryos to genetically “vaccinate” against HIV infection. It has argued that there should be an initial period during which no clinical use of germline editing is allowed at all. Research would still be allowed, provided embryos are used only in the very early stages in laboratory studies, and not transferred to a woman’s uterus to develop further and this period could last five years.

**Conclusions:** We conclude that ethical rules can change with every development. Currently, scientists think that ethical justifications have to be confirmed.

**Keywords:** morality, crispr-cas9, gene edited babies

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## A Beta-Lactamase Inhibitor Identified Rough Virtual Screening

*İklima ZÜNBÜL, Ebru ÇAKA, Yusuf Ceyhan ERDOĞAN, Duygu ŞENTÜRK, Michaela GULEA, Onur SERCİNOĞLU, Temra ÖZBEK, Berna SARIYAR AKBULUT*

$\beta$ -lactams are well-known and widely used antibiotics with  $\beta$ -lactam rings in their structure. They block the cell wall synthesis by binding the bacterial transpeptidases. This disrupts the cell wall and bacteria eventually die. Misuse of  $\beta$ -lactam antibiotics has forced bacteria to develop different resistance strategies. Among these,  $\beta$ -lactamase production is the predominant mechanism. These enzymes bind the amide bond of the  $\beta$ -lactam ring and thus render the antibiotic ineffective. The current work undertakes the effort to find a  $\beta$ -lactam inhibitor that targets the allosteric site of the enzyme. Taking TEM-1  $\beta$ -lactamase (PDB code: 1PZO) as the target MuTaLig-generated chemical library Chemothea was screened for the best binders.

Virtual screening was performed with AutoDock Vina 1.1.23 and then binding energies were estimated. The first 100 binders were taken and further refined by screening their ADME properties using DruLiTo, which refined the results to 32 binders. Among these, we were able to test only 3 molecules to test and found CM840 with the potential to inhibit  $\beta$ -lactamase. The authors acknowledge The Scientific and Technological Research Council of Turkey (TUBITAK-118Z572)

Keywords: beta lactamase, virtual screening, inhibitor

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## **Potential Probiotic Activity of Marine Seaweed: “Spirulina” by Using Novel Enzyme Endo-Beta-N-Acetylglucosaminidase**

*Damla YAY, Dilfuza ERNAFASOVA, Merve KAPLAN, Secan KARAV, Hacı Mehmet KAYILI*

Marine oligosaccharides have attracted increasing attention recently in developing potential drugs, as extraordinary resources for the discovery of functional foods, pharmaceuticals, and biomaterials. Such targeted species include blue-green microalgae *Spirulina* sp. *Spirulina* sp. include a high variety of glycoproteins which have important biological functions. As known, glycoproteins include glycan /oligosaccharides and protein part and glycans can be attached to the protein by O- OR N- glycosidic bonds via a process called glycosylation. Glycosylation is one of the most common and important post-translational modification of proteins that give them biological activity. Actually, by releasing these conjugated glycans we could understand the function of glycans on these glycoproteins and at that point we used a novel glycosidase EndoBI-1 isolated from an infant microbe is capable of cleavage of N type glycan core of a wide variety of proteins. Free glycans regarded as prebiotic that allows selective growth of probiotics and they have high impact and quite beneficial effect on the human gut microbiota. This research will help us to understand the glycan profile of *Spirulina* oligosaccharides and shed light on how to shape our intestinal microflora with increasing consumption.

Keywords: prebiotic, glycan, glycosylation, algae

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# Multidisciplinary Analyses of NF-1 Mutated Scoliosis Patient Recognized for the First Time

Onur SÜER, Özden BEDRE, Hüseyin Onay, Gökhan GÖKMEN, Figen GÖVSA

Background: Scoliosis may be accompanied by other genetic diseases.

Aim: This study was to analyzed a patient who treated at the orthopaedics clinic and who followed up by digital analysis method in 100 scoliosis patients with genetic analysis. Distance between anatomic points and angles were calculated by Image J software program and thoracolumbal curvature was measured by direct radiogram using Cobb method. In a patient, NF1 region screening was analyzed by amplifying 48 parts between exons 1-58.

Results: Cervical lordosis angle was 15.20, thoracic kyphosis angle was 57.40 and lumbar lordosis angle was 21.60. A positive correlation was found between thoracic kyphosis angle obtained by photoanthropometric analysis and thoracic curvature degree obtained by Cobb method ( $r=0,541$ ). NF1 mutation screening report heterozygous c.3052\_3056delTTAGT mutation was detected. This is a case not previously detected in any database.

Conclusion: Analysis with the Mutation taster modeling program suggests that this mutation may impair protein function. It is thought that this change may be related to NF Type 1, McCune Albright syndrome. Only mutations in exons and exon-intron components are evaluated in the analysis.

Keywords: NF type 1, McCune-Albright Syndrome, scoliosis, photo-anthropometry, spine anatomy.

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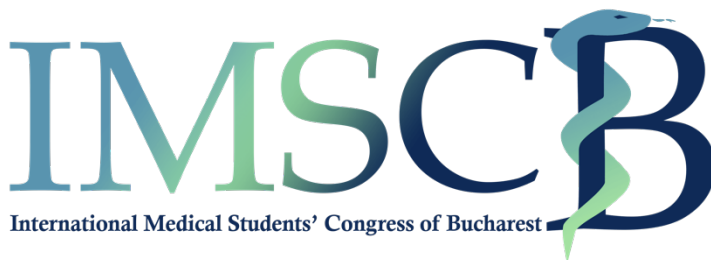


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## NOTES

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