Clinical Medicine
Prophylactic Medicine
Theoretical Medicine
Stomatology & Dentistry
Veterinary Medicine and Zoo
Drug Technology and Organization of Pharmaceutical Business
Pharmaceutical Chemistry and Pharmacology
Standardization and Organization of Medicines Production
History of Pharmacy
Innovations in Medicine
Biophysics and Biochemistry
Radiology and Microbiology
Molecular Biology and Genetics
Botany and Virology
Microbiology and Hydrobiology
Physiology of Plants, Animals and Humans
Ecology, Immunology and Biotechnology
Virology and Immunology
History of Biology
Entomology

http://sc-media.org/ambiance-in-life-isjmsc/
Always laugh when you can. It is cheap medicine.    Lord Byron

03.01.2020     ISSN: 2346-8068; E-ISSN: 2346-8181; DOI: 10.36962/ALISJMSC

© SOUTHERN CAUCASUS SCIENTIFIC JOURNALS

AMBIANCE
INTERNATIONAL SCIENTIFIC JOURNAL IN MEDICINE
MULTIDISCIPLINARY JOURNAL
REFEREED & REVIEWED JOURNAL

JOURNAL INDEXING

General Impact Factor  2017 – 2.3892

GEORGIA, TBILISI 2020
Editors-in-chief:

Managing Director and Editor-in-chief: Tamar Didbaridze

Editor: Eter Buknikashvili

EDITORIAL BOARD LIST SEE PAGE 40

ISSN: 2346-8068; E-ISSN: 2346-8181; DOI PREFIX: 10.36962 / ALISJMSC

©Publisher: LTD International Research, Education & Training Center. (UK, London),
Director and shareholder: Alexandra Cuco, Lawyer. Portugal.
Deputy and shareholder: Namig Isazade, PhD in Business Administration.
©Editorial office: 71-75 Shelton Street, Covent Garden, London, WC2H 9JQ, UK.
Registered address: 71-75 Shelton Street, Covent Garden, London, WC2H 9JQ, UK.
Telephones: +994 55 241 70 12; +994 51 864 88 94
Website: http://sc-media.org/
E-mail: gulustanbssjar@gmail.com, sc.mediagroup2017@gmail.com

©Publisher: NGO International Research, Education & Training Center.
Deputy and founder of organization: Seyfulla Isayev. Azerbaijan Marine Academy.
©Typography: NGO International Research, Education & Training Center. BS Journals.
Registered address: Narva mnt 5, 10117 Tallinn, Estonia.
Telephones: +994 55 241 70 12; +994518648894; +994 55 241 70 09
Website: http://sc-media.org/
E-mail: gulustanbssjar@gmail.com, sc.mediagroup2017@gmail.com, Caucasusblacksea@gmail.com

©Publisher: Representation of Azerbaijan International Diaspora Center in Georgia. SCSJAR.
©Typography: Representation of Azerbaijan International Diaspora Center in Georgia. SCSJAR.
Registered address: 0165 Georgia. Marneuli municipality. Village Takalo.
Telephones: +994 55 241 70 09; +994 55 241 70 12; +995 59 201 66 14
Website: http://sc-media.org/
E-mail: gulustanbssjar@gmail.com, Caucasusblacksea@gmail.com

© The Southern Caucasus Media. NGO RAIDCG, MTÜ IRETC. All rights reserved. Reproduction, store in a retrieval system, or transmitted in any form, electronic, mechanic photocopying of any publishing of Southern Caucasus Scientific Journals permitted only with the agreement of the publisher. The editorial board does not bear any responsibility for the contents of advertisements and papers. The editorial board’s views can differ from the author’s opinion. The journal published and issued by The Southern Caucasus Media.
### TABLE OF CONTENTS

<table>
<thead>
<tr>
<th>Author(s)</th>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Irine Ioramashvili, Rusudan Sujashvili, Marika Gamkrelidze, Sofia Tsitsilashvili</td>
<td>Sensitivity of Serum Proteins of GI Cancer Patients to Chemotherapy Courses</td>
<td>05</td>
</tr>
<tr>
<td>Melano Shavgulidze, Eka Chkhartishvili, Mariam Babilodze, Nino Rogava, Nargiz Nachkebia</td>
<td>Changes in Open Field Behavior and Declerative Memory in “Depressive” Rats with High Immobility and Decreased Level of Brain Monoamines</td>
<td>06</td>
</tr>
<tr>
<td>Gulmira Zharmakhanova, Lyazzat Syrlybayeva, Eleonora Nurbaulina, Lyazzat Baikadamova, Gulaiym Eshtayeva</td>
<td>Preliminary Results on the Using Tandem Mass Spectrometry in Diagnosis of Inherited Metabolic Diseases</td>
<td>08</td>
</tr>
<tr>
<td>Afig Berdeli</td>
<td>Development of New Genetic Approaches and Their Application in the Diagnosis of Mendeliome Diseases</td>
<td>10</td>
</tr>
<tr>
<td>Esra Cholak Genish, Fethi Sirri Cham</td>
<td>A Rare Case with Cleft Palate / Lip: Partial Trisomy 8q24</td>
<td>13</td>
</tr>
<tr>
<td>Aytan Mammadbayli, Sona Aliyeva, Madina Taghiyeva</td>
<td>Cerebellar Ataxia and Seizures in Patient with Coenzyme Q Deficiency</td>
<td>14</td>
</tr>
<tr>
<td>Mustafa Salikhov, Sona Shakhbazbekova, Ilaha Rasul, Inara Alizade</td>
<td>Criterion of Diagnosis of Heart Damage in Compensated Cirrhosis of the Liver in the Practice of Family Doctor</td>
<td>15</td>
</tr>
<tr>
<td>Aytakin Hasanova</td>
<td>Molecular Genetic Diagnostics of Chromosomal Diseases with Multiplex Ligation-Dependent Probe Amplification</td>
<td>17</td>
</tr>
<tr>
<td>Aytakin Hasanova, Gulnara Kuliyeva</td>
<td>The Frequency and Distribution of Sister Chromatid Exchanges (Sces) In the Individual Chromosomes of Human Karyotype</td>
<td>18</td>
</tr>
<tr>
<td>Aytakin Hasanova, Simuzar Hajizada</td>
<td>Working Conditions and Health of Seafarers</td>
<td>20</td>
</tr>
<tr>
<td>Mustafa Salikhov, Kamile Salikhova</td>
<td>Family Doctor and Medical Genetics</td>
<td>22</td>
</tr>
<tr>
<td>Aynur Panahova</td>
<td>The Importance of Electron Microscopic Systematic Classification We Offer in Cervical Cancer</td>
<td>23</td>
</tr>
</tbody>
</table>
Zarintaj Rustamova, Lala Akhundova, Gulmira Alibayova, Nurmammad Mustafayev, Irada Huseynova
ASSOCIATION OF RS7903146 C/T POLYMORPHISM OF TCF7L2 GENE WITH TYPE 2 DIABETES MELLITUS IN AZERBAIJAN POPULATION: PRELIMINARY STUDIES .......................................................... 25

Narmin Salayeva
MOLECULAR GENETIC ANALYSIS OF GLA GENE CAUSING INHERITED FABRY DISEASE FOR POPULATION OF AZERBAIJAN REPUBLIC................................................................................................. 26

Aysel Hashimova
HUMAN AS A BIOTIC FACTOR IN ACCUMULATION OF MICROELEMENTS WITH WEAK TOXICITY IN BONE AND MUSCLE TISSUE OF BACKGROUND REPTILE SPECIES IN THE ABSHERON PENINSULA .......................................................... 28

Gulnara Nasrullahyeva, Vafa Mammadova, Gunay Aliyeva, Elnura Atakishiyeva
IMMUNOLOGICAL AND GENETIC ASPECTS OF HEREDITARY ANTIBODY DEFICIENCIES ..................... 31

Ziba Nasibova
EARLY EPILEPTIC ENCEPHALOPATHY GENETICS OF PATIENT IN AZERBAIJAN ...................................... 33

Tahira Askarova
DIFFERENT GENETIC FORMS OF HEREDITARY HEMOCHROMATOSIS IN AZERBAIJAN POPULATION .......................................................... 36

Nargiz Garayeva
THE ROLE OF ANEUPLOIDY IN THE UPREGULATION OF E2F4 AND E2F6 GENES IN BREAST CANCER CELL LINES .................................................................................................................. 38

Lala Allahverdiyeva, Naile Guliyeva
İMMUNE DİAGNOSİS OF CANDLE-LIKE SYNDROME, AN AUTO-INFLAMMATORY DİSEASE ................. 39
SENSITIVITY OF SERUM PROTEINS OF GI CANCER PATIENTS TO CHEMOTHERAPY COURSES

1Irine Ioramashvili, 2Rusudan Sujashvili, 3Marika Gamkrelidze, 4Sofia Tsitsilashvili

1,2,3,4 Iv.Beritashvili Center of Experimental Biomedicine, New Vision University.

ABSTRACT

Gastrointestinal cancers (GI) are one of the most abundant types of cancers among the world population, though statistical data indicate that in eastern Asia these types of cancer occur 4 times more often than in Western Europe. Absence of treatment of bacterial infections, obesity, and lack of vegetable food in a diet can be the case of GI cancer. All pathologies are inevitably connected to the changes in cell cycle, abnormal protein amount and their dysfunction. Serum proteins are widely used as an additional source of information about body condition, also changes in protein composition can point out the mechanism of disease development and effectiveness of treatment. In the presented work we studied protein composition of GI cancer patients in different stages of cancer development, after and before chemotherapy and compared these data to protein composition of healthy control group of voluntaries. Treatment of patients was performed according the guidelines appropriate for the GI cancer. Association of the effectiveness of treatment at the different stages of chemotherapeutic courses and changes of protein composition of blood serum has been assessed. Proteins composition was studies by SDS-PAGE electrophoresis and densitometry analysis. Experimentally gained molecular and statistical information exposed the most vulnerable groups of proteins affected by chemotherapeutic agents indicating targets for searching new biomarkers for treatment effectiveness.

Research involving human patients performed in accordance with the requirements of the Council of Europe Convention on Human Rights and Biomedicine, Biomedical Research, as well as the UNESCO Declaration of Bioethics and Human Rights.

Key wards: Gastrointestinal cancer, chemotherapy, proteins, biomarkers.

REFERENCES

1. R.Sujashvili, Tsitsilashvili S., M.Abuladze, “Elisa test of changes of serum ubiquitin levels in GI cancer patients at the different stages of treatment” International Conference “Cancer Metastasis”, Seefeld-in-Tirol, Austria; December 11 – 14, 2019;
Changes in open field behavior and declarative memory in “depressive” rats with high immobility and decreased level of brain monoamines

Melano Shavgulidze, Eka Chkhartishvili, Mariam Babilodze, Nino Rogava, Nargiz Nachkebia

Lab Neurobiology of sleep-wakefulness cycle, I. Beritashvili Center of experimental Biomedicine. (Georgia)

Email: meko_shavgulidze@yahoo.com

Introduction
Changes in some forms of motivational-emotional behavior, learning and memory are thought to be characteristic for major depressive disease. However, results existing until today about the character of changes in motivational-emotional and exploratory behavior as well as character of disorders in declarative memory, accompanying major depressive disease, are not unambiguous. Therefore, studying them in animal models of depression is very topical and important.

Methods
Experiments were conducted on adult white wild rats (with 250-300 g weight). “Depressive” and “non-depressive” rats were selected according to the level of immobility in forced swim test. Rats with low level of immobility, “non-depressive” rats, constituted control group and rats with high level of immobility, “depressive” rats, constituted the experimental group (10 rats in each).

Changes of motivational-emotional and exploratory behavior were studied in open field test. The changes of learning and memory were studied in the fear motivated one trial passive avoidance test considered as the declarative memory test. Experiments were carried out on “non-depressive”, control and “depressive”, experimental groups (10 rats in each).

Obtained results were processed statistically by Student’s t-test.

Results
Sharp decrease in locomotion was found in rats with high level of immobility. It was manifested in a significant decrease of the number of crossed squares. The quantitative indices of vertical activity, vertical standings, head risings, were also sharply decreased. Fear reaction was considerably increased in “depressive” rats, manifested in the significant decrease of the number of entering in the center of open field and grooming and sharp increase in defecation rate.

Investigation of the changes of learning and memory in the passive avoidance test has shown that the latency of entering from the light into dark section of passive avoidance camera, in the learning session, was sharply increased in “depressive” rats. They revealed an impaired ability to evaluate the level of danger coming from the brightly illuminated open area and therefore they do not hurry to escape from the dangerous section. The difference between “depressive” and “non-depressive” rats was maintained even after 24 hours from receiving a painful stimulation. In particular, the animals of control group remember that they have received a painful stimulation in dark section during learning session and do not enter there during testing session, whereas the experimental animals with considerable delay but still enter in the dark section during testing session, therefore, they show significant impairment of declarative memory in passive avoidance task.

Conclusions
Locomotor and exploratory behavior are impaired and fear motivation is increased in the open field in “depressive” rats with high immobility and low level of monoamines content in the brain. Learning and memory in one of the tests of declarative memory, so called passive avoidance task, is disturbed.

Keywords: “Depressive” rats, Open field Behavior, Declarative Memory and Monoamines Deficiency.

References
4. R. Sujashvili, Tsitsilashvili S., M. Abuladze, “Elisa test of changes of serum ubiquitin levels in GI cancer patients at the different stages of treatment” International Conference “Cancer Metastasis”, Seefeld-in-Tirol, Austria; December 11 – 14, 2019;
PRELIMINARY RESULTS ON THE USING TANDEM MASS SPECTROMETRY IN DIAGNOSIS OF INHERITED METABOLIC DISEASES

Gulmira Zharmakhanova¹, Lyazzat Syrlybayeva², Eleonora Nurbaulina³, Lyazzat Baikadamova⁴,
Gulaiym Eshtayeva⁵

¹Head of the department of molecular biology and medical genetics, West Kazakhstan Marat Ospanov Medical University, Aktobe, Kazakhstan,
²Senior lecturer of the department of molecular biology and medical genetics, West Kazakhstan Marat Ospanov Medical University, Aktobe, Kazakhstan,
³Senior lecturer of the department of general medical practice, West Kazakhstan Marat Ospanov Medical University, Aktobe, Kazakhstan,
⁴Obstetrician-gynecologist, Medical Center Aktobe, Kazakhstan,
⁵Senior lecturer, Higher Medical college, Almaty, Kazakhstan

BACKGROUND
According to the results of the researches common indexes of the prevalence of inherited metabolic diseases (IMD) varies from 1 to 800 on 1 to 2500 alive newborns. IMD are taking one of the first places among children pathology, early children death (40%) and disability[1]. According to systematic review of the 43 forms of the inborn errors of metabolism are related to unexpected death of newborns. For IMD it is common to have a wide spectrum of the unusual clinical manifestation, often they are not diagnosed, while well timed diagnoses and proper treatment are able to prevent severe systematic lesions, which lead to death and disability[2]. For that reason one of the most significant problems of the modern pediatrics is to early diagnosis of IMD. The only way to diagnosis of orphan metabolic diseases is the tandem mass spectrometry (TMS) [3].

AIM
Scientifically substantiate the need for implementation of selective screening IMD of children using TMS method in Republic of Kazakhstan (RK) for early diagnosis, therapy of the inherited metabolic diseases, to reduce disability and death rate.

MATERIALS AND METHODS
Material of the research – dry blood spots, taken using standard methodology on filtered DBS papers, which are used in RK in the program of neonatal screening (for retrospective research – archived samples of the dry blood spots of the children dead during first year of life). Method of the research is tandem mass spectometry (QSight Perkin Elmer).

RESULTS
Analysis of the archived dry blood spot samples showed metabolic deviations in 20.4% of the cases. The detected changes are related to amino acids metabolic disorders, defects of β-oxidation of the fat acids, decrease activity of the glucocerebrosidase (Gaucher’s disease) and sphingomyelinase (Nimman – Pick disease). Results of the selective screening have shown metabolic disorders in 5% of the cases (defects of β-oxidation of the fat acids, aminoacidopathy, organic aciduria).

CONCLUSIONS
The preliminary results of the using TMS for the diagnosis of IMD have shown the need for implementation of selective screening IMD using TMS, which is able to conduct diagnosis of 75 metabolites of 49 IMD in single blood spot, which were not detected in RK previously. Taking into the consideration economic expenses of the government, related to the costs of the systematic treatment, medical service, life expectancy and lifelong support of the disabled children with IMD, early detection of orphan metabolic diseases is the vital condition of the decrease of newborn and children death rate, sickness rate and disability.

This research study was carried out as a part of a scientific project funded by West Kazakhstan Marat Ospanov Medical University.

REFERENCE

2. Е.А. Николаева, «Значение достижений медицинской генетики для решения проблемы нарушения...

THE FIRST INTERNATIONAL SCIENTIFIC PRACTICAL VIRTUAL CONFERENCE HUMAN GENETICS AND GENETIC DISEASES: PROBLEMS AND DEVELOPMENT PERSPECTIVES
развития у детей»// Российский вестник перинатологии и педиатрии, 2, 2016 г., с.5-10
DEVELOPMENT OF NEW GENETIC APPROACHES AND THEIR APPLICATION IN THE DIAGNOSIS OF MENDELIOME DISEASES

Afig Berdeli
Professor, PhD MD.
Ege University Faculty of Medicine, Department of Pediatrics, Molecular Medicine Laboratory. (Turkey)

Since the existence of human kind, they wondered what was happening around them, tried to understand and tried to overcome the difficulties they faced by doing new discoveries. Supportive and complementary researches carried out by scientists from past to present form the basis of today's technologies. In 1977, two different DNA sequence analysis methods were found by Allan MAXAM- Walter GILBERT and Frederick SANGER. In 1982, Akiyoshi proposed automatic analysis of DNA sequence and robots have begun to be developed. These types of studies have brought up the sequencing of the entire human genome. The Human Genome Project was a 13-year-long, publicly funded project initiated in 1990 with the objective of determining the DNA sequence of the entire euchromatic human genome within 15 years. In its early days, the Human Genome Project was met with skepticism by many people, including scientists and nonscientists alike. One prominent question was whether the huge cost of the project would outweigh the potential benefits. Today, however, the overwhelming success of the Human Genome Project is readily apparent. Not only did the completion of this project usher in a new era in medicine, but it also led to significant advances in the types of technology used to sequence DNA. The Human Genome Project, the mapping of our 30 000 genes and the sequencing of all of our DNA, will have major impact on biomedical research and the whole of therapeutic and preventive health care. The purpose of the human genome project is to find genes and describe what they do. This is an important distinction from some of the previous approaches to genetics because the aim is not to define function at this stage but to focus on mapping the genes and clarifying their sequence. It was announced that the participants of the Human Genome Project and Celera completed the draft of the human gene map in June 2000.

A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Genetic disorders can be caused by a mutation in one gene (monogenic disorder) or by mutations in multiple genes (genetic heterogeneity). Genetic Diagnostic testing is used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms. Diagnostic testing can be performed before birth or at any time during a person's life, but is not available for all genes or all genetic conditions. The results of a diagnostic test can influence a person's choices about health care and the management of the disorder. In the years 2000 and thereafter different molecular methods have been used in the diagnosis of monogenic diseases with Mendel inheritance (Mendeliome.) Among these methods, Sanger sequencing method has been accepted as the gold standard which is the most frequently applied and allows to read the entire sequence of genes. Molecular Medicine Laboratory, which I have managed since 2001, has molecular diagnosis of 512 different genetic diseases in 42000 patients. Over the years, this approach has led to answer some of our questions in the group of diseases or syndromes that show genetic heterogeneity (proteinuria, cystic kidney diseases, collagen diseases, autoinflammatory diseases, etc.). In these years, new DNA sequencing methods have been developed and these have led to the development of new technologies in genetics. The emergence of next generation sequencing (NGS) techniques has made the sequencing of whole genomes, transcriptomes, and epigenomes faster and more readily available than previous methods such as Sanger sequencing, which was developed in the 1970s. The commercialisation of next generation sequencing (NGS), approximately 10 years ago, led to major improvements in both research and the diagnosis of genetic diseases. NGS makes it possible to sequence the whole genome, exome, or predetermined panel of a patient’s genes in a single sequencing reaction and in a much more time efficient manner. NGS also has the capacity to characterize all steps of transcription, translation, and methylation of DNA.

Next generation sequencing (NGS), massively parallel or deep sequencing are related terms that describe a DNA sequencing technology which has revolutionized genomic research. Using NGS an entire human genome can be sequenced within a single day. In contrast, the previous Sanger sequencing technology, used to decipher the human genome, required over a decade to deliver the final draft. Although in genome research NGS has mostly superseded conventional Sanger sequencing, it has not yet translated into routine clinical practice. The National Human Genome Research Institute subsequently initiated and funded a sequencing technology development program with the aim of reducing the duration and cost of genome sequencing. This led to a wave of new projects, and finally to the introduction of the contemporary commercial sequencing platforms. NGS permits the simultaneous analysis of multiple genetic aberrations, including single nucleotide variants (SNVs), small insertions/deletions (indels) as well as copy number variants (CNVs) or complex genomic rearrangements. Although nowadays sequencing-by-synthesis is the predominant sequencing technology in use, multiple technologies and platforms have been developed and are
commercially available. The scope of available strategies for genome sequencing ranges from targeted gene panels encompassing several thousand base calls through whole-exome sequencing (WES) analysis of the ~22,000 human protein-coding genes (40–50 million bases) to WGS across all 3.3 billion bases of the human genome. NGS found its first clinical application in germline testing for known monogenic and rare diseases by targeted panels, while it was shown that WES is ideally suited for the diagnosis of suspected novel Mendelian diseases.

We are studying on hereditary autoinflammatory disease panels in our molecular medicine laboratory with the NGS method. The monogenic autoinflammatory diseases are a group of illnesses with prominent rheumatic manifestations that are characterized by genetically-determined recurrent sterile inflammation, and are thus inborn errors of innate immunity. These are hereditary autoinflammatory diseases analysis the mutations of FMF, HIDS, TRAPS, PFAPA, DIRA, Majeed Syndrome, CRMO, PAPA, Schnitzler Syndrome Blau (NOD2) (PGA), NLPR12, CANDLE syndrome, Behcet’s, SJIA, CAPS, FCAS2, FCAS3, FCAS4, interferonopathies and ubiquitination disorders.

More than 3000 patients was analyzed with AID targeted NGS panel and genetic diagnosis was made in more than 60% of patients. 1200 patients were analyzed with nephrotic syndrome panel consisting of 29 genes and molecular classification was performed in 29% of patients. Atypical HUS panel consisting of 12 genes was made in 400 patients and 111 variants were detected in these patients. Of these, 58 (52%) were identified in our study, and 53 (48%) were previously reported in the HGMD database. In our Alport disease panel, we studied COL4A3, COL4A4, COL4A5 genes on 130 patients. We detected 61 different variant on these patients. 9 of the total 61 variants evaluated are the new mutations detected in this study. 21 of all mutations detected were classified as benign, 3 as possible pathogens and 28 as pathogens according to bioinformatics databases. In this study, in addition to the mutations detected in the literature, 9 new mutations were detected. PKD1, PKD2 and PKHD1 genes which responsible for polycystic kidney disease were analyzed in 190 patients by targeted NGS method. There were 134 missense mutations and 18 nonsense mutations in the patients. 35 of them are new mutations detected in this study. 142 mutations reported in these genes in the literature were also detected in this study. Our renal channel disease panel consist of 17 genes and urolithiasis panel consist of 30 genes. We use these panel on routine laboratory tests.

In conclusion, Sanger genetic mutation analysis method is the gold standard in single gene diseases that molecular pathogenesis is well known. It’s important to confirmation in clinical differential diagnosis and establishing new diagnoses in single gene diseases. This method is important in the re-molecular classification of genetic diseases, in monitoring and managing treatment, in planning prognosis, in the emergence of target therapies, in reproductive genetic consultations, and in rapid predictive genetic consultations in family members both symptomatic and non-symptomatic. NGS, the new technological approach, should be applied in kidney diseases which phenotype has genetic heterogeneity. Mutation classifications and bioinformatics data should be used wisely, and accurate interpretation of NGS findings requires a multidisciplinary approach, often involving specialist physicians, clinical geneticists and laboratory specialists.

REFERENCES

2. A M Maxam ; W Gilbert, A new method for sequencing DNA. PNAS, 1977 74 (2) 560-564
A RARE CASE WITH CLEFT PALATE / LIP: PARTIAL TRISOMY 8Q24

1Esra Cholak Genish, 2Fethi Sirri Cham
1,2Manisa Celal Bayar University Faculty of Medicine, medical genetics, Manisa (Turkey)

ABSTRACT

OBJECTIVES
Cleft palate / lip is one of the common congenital craniofacial anomalies and its prevalence is 1-2 / 1000. This anomaly requires a multidisciplinary approach because it is important in the physical and psychological development of the patients. Cleft palate / lip may be accompanied by a syndrome or may be seen as isolated. In this study, we aimed to present a rare partial trisomy 8 in a syndromic patient with cleft palate / lip and accompanying congenital anomalies occurring as a result of maternal balanced translocation.

MATERIAL AND METHODS
Peripheral venous blood chromosome analysis was performed on proband and the parents’. Phytohemagglutinin (PHA) - induced peripheral blood lymphocytes cultures were used for the study. The chromosomes of 30 G-banded metaphases (500-550 band level) were examined for numerical and structural chromosome abnormalities. The thirty metaphase areas stained with giemsa trypsin banding technique were evaluated according to the 2016 International System for Human Cytogenetic Nomenclature (ISCN).

RESULTS
On the physical examination of the newborn, cleft palate / lips and umbilical hernia were detected. There was no consanguineous marriage between his parents. Chromosome analysis showed that the mother had a balanced reciprocal translocation carrier, 46, XX, t (8; 17) (q24.1; p13). Patient 46, XY, der (17), t (8; 17) (q24.1; p17) mat chromosome was detected.

CONCLUSIONS
Conventional karyotyping analysis revealed a rare trisomy 8q24 syndrome. Chromosomal anomalies are a condition that must be kept in mind with various findings from prenatal period to adulthood. Individuals with a balanced chromosomal translocation carrier may transmit this condition unsteadily to children with abnormal segregation during gametogenesis. This balanced translocation, which is also found in the mother, has clinical findings consistent with trisomy 8q24 syndrome, which is a rare disorder resulting in unbalanced transfer of the patient.

Keywords: Cleft lip and palate (CL / P), reciprocal translocation, trisomy 8q24.

REFERENCES

CEREBELLAR ATAXIA AND SEIZURES IN PATIENT WITH COENZYME Q DEFICIENCY

Aytan Mammadbayli, Sona Aliyeva, Madina Taghiyeva

Department of Neurology, Professor. Department of Neurology. Child Neurology Hospital. MD. Department of Neurology. MD. Doctoral candidate.

Azerbaijan Medical University. (Azerbaijan)

ABSTRACT

OBJECTIVE

Primary coenzyme Q(10) deficiency represents a clinically heterogeneous condition suggestive of genetic heterogeneity, and several disease genes have been previously identified. These patients presented a similar progressive neurological disorder with cerebellar atrophy and seizures. Cerebellar ataxia is a common symptom of coenzyme Q10 (CoQ10) deficiency associated with COQ8A mutations.

METHODS

The patient is a boy born at term, parents are cousins. Pregnancy, birth history and developmental milestones were unremarkable. Before neurological manifestation, they had night recurrent vomiting(1-2 times per month) not associated with food intake. He developed his first focal seizure at 9 years. Since that time, he continued to had focal and generalized seizures which were responsive to levetiracetam and also had epileptic status. Neurologically, he was noted to have additional symptoms after epileptic status that included left sided spastic hemiparesis, facial left sided palsy, positive oral automatism reflexes, positive pathological reflexes in the left, clonus in both legs and cerebellar symptoms (ataxia—dynamic and static, slurred speech, intention tremor). His cognitive functions were good before epileptic status. Metabolic workup revealed persistently elevated lactate and low levels of pyruvate. Magnetic resonance imaging (MRI) of the brain was normal. EEG shows focal epileptic activity of the occipital lobe.

RESULTS

Using whole exome sequencing, we identified compound heterozygous variants in the COQ8A gene p.R271L/p. L506W heterozygous mutation was identified.

CONCLUSIONS

This patient presenting with seizures and cerebellar symptoms (ataxia—dynamic and static, slurred speech, intention tremor) represents rare mitochondrial disease caused by biallelic COQ8A mutations. The response to CoQ10 supplement is good and patient remains stable.

Keywords: coenzyme Q10, COQ8A, cerebellar ataxia, seizures.

REFERENCE

CRITERION OF DIAGNOSIS OF HEART DAMAGE IN COMPENSATED CIRRHOSIS OF THE LIVER IN THE PRACTICE OF FAMILY DOCTOR

Mustafa Salikhov, Sona Shakhbazbekova, Ilaha Rasul, Inara Alizade

1Head of the Department of Family Medicine, Associate Professor.
2Department of Family Medicine, Associate Professor.
3Assistant of the Department of Family Medicine.
4Assistant of the Department of Family Medicine.
1, 2, 3, 4Azerbaijan Medical University.

ABSTRACT

With the intention of seeking preclinical clinical manifestations of heart damage we examined 34 patients aged 40-45 years with compensated cirrhosis of the liver in an outpatient setting. In 65% of patients determined hypertrophy and dilatation of left atrium, counting diametrical and longitudinal measurement, and also disturbance of diastolic function of left ventricule. In order to identify heart damage in patients with compensated cirrhosis of the liver it is necessary carrying out ECG and doppler echocardiography, which allow at early stage to identify damage of the heart muscle and timely correct disfunction of the heart, which in turn prevents transition of cirrhosis of the liver to the decompensatory stage.

TARGET OF RESEARCH
to search preclinical manifestations of heart damage in patients with compensated cirrhosis of the liver in out-patient condition.

METHODS
We have examined 34 patients with cirrhosis of the liver, age 40-45 years, who were registered in the polyclinic. All patients have been carried out liver function tests, also fasting blood glucose, levels of albumin, creatinine and lipid profile. Criterion of exclusion was availability of decompensated cirrhosis. Retained 30 patients with compensated cirrhosis performed ECG, taking into consideration prolongation of QT interval on Bazetts formula and doppler echocardiography.

RESULTS
Echocardiography revealed -20 patients have sinus tachycardia, 10% have left bundle brunch block and ventricular extrasystoles; average duration of QT interval -40.0±8.2 mc. Echocardiography revealed ejection fraction at rest within normal limits (62.4±6.1%), increased density of ventricular septum and back wall of left ventricule 0.91±0.9 sm and 0.98±0.12 sm accordingly. In 65% of patients determined hypertrophy and dilatation of left atrium, counting diametrical and longitudinal measurement, and also disturbance of diastolic function of left ventricule (dilatation of left atrium up to 40 mm and increase of diastolic pressure in left ventricule).

CONCLUSION
In order to identify heart damage in patients with compensated cirrhosis of the liver it is necessary carrying out ECG and doppler echocardiography, which allow at early stage to identify damage of the heart muscle and timely correct disfunction of the heart, which in turn prevents transition of cirrhosis of the liver to the decompensatory stage.

REFERENCES
1. Cirrhosis: Diagnosis and Management. Andrew Smith, MD; Katrina Baumgartner, MD; and Christopher Bositis, MD, Greater Lawrence Family Health Center 2019
2. EASL Clinical Practice Guidelines for the management of patients with decompensated cirrhosis 2018
3. Современные принципы диагностики и лечения осложнений цирроза печени, Москва 2018
DNMT3B -579G>T POLYMORPHISM AND THE RISK OF COLORECTAL CANCER IN AZERBAIJAN POPULATION

Bayram Bayramov¹, Khagani Eynullazada², Nigar Karimova³, Hazi Aslanov⁴, Vugar Yagublu⁵, Nuru Bayramov⁶

¹,²,³Genetic Resources Institute of Azerbaijan National Academy of Sciences. (Azerbaijan)
⁴Scientific Surgical Center named after Academician Topchubashov. (Azerbaijan)
⁵Hospital Frankfurt Hoechst, Frankfurt am Main. (Germany)
⁶Department of surgical diseases I, Azerbaijan Medical University.

BACKGROUND
DNA methylation is one of the important mechanisms for epigenetic modifications and under the control of DNA methyltransferases (DNMTs). The family of DNMTs consists of five different enzymes (DNMT1, DNMT2, DNMT3A, DNMT3B, and DNMT3L). The purpose of this case-control study was to determine the association between DNMT3B -579 G>T polymorphism and the risk of colorectal cancer in the Azerbaijan population.

MATERIALS AND METHODS
Blood samples were collected from the Center of Scientific Surgery Named After Academician Topchubashov. One hundred and fifteen patients with colorectal cancers and 115 controls were included in the study. Genomic DNA isolation was performed with the Qiagen kit (QIAamp DNA Mini Kit) in the Human Genetics Laboratories of the Institute of Genetic Resources. The genotyping of DNMT3B -579G>T was performed by PCR-RFLP.

RESULTS
The frequencies of the GG, GT, and TT genotypes of DNMT3B were 41.7%, 40%, and 18.3% in the patients with colorectal cancer and 28.7%, 54.8%, and 16.5% in the healthy control group respectively. The GT genotype was significantly different between patients with colorectal cancer and control subjects. Mutant TT genotype was more frequent in the patient group compared to the control group.

CONCLUSION
Our findings suggest that DNMT3B -579G>T polymorphism represents a genetic risk factor that may play an important role in colorectal cancer development.

Keywords: DNA Methyltransferase; Genes; Colorectal; Cancer; Polymorphism

REFERENCES
MOLECULAR GENETIC DIAGNOSTICS OF CHROMOSOMAL DISEASES WITH MULTIPLEX LIGATION-DEPENDENT PROBE AMPLIFICATION

Aytakin Hasanova

Azerbaijan Medical University, Department of Medical Biology and Genetics, Azerbaijan

ABSTRACT

Chromosomal diseases are birth defects caused by changes in the number or structure of chromosomes. Among newborn children, the frequency of chromosomal pathology is up to 1.0%. Trisomy of 21 chromosomes, which leads to Down syndrome, is most common. Polysomies on X and Y chromosomes are also noted, with an abnormal number of sex chromosomes often showing themselves only at pubertic age. Changes in the structure of chromosomes (aberrations) are less common, but cause profound disorders in the development of many organ systems. The most severe clinical picture is seen in the deletion (loss) of part of the chromosome, it may be the deletion an entire shoulder or only a chromosome locus. Lack of genetic material leads to severe defects. The duplication (doubling) of the chromosome section may, among other things, affect the mental and psychological development of the patient, but usually does not lead to pronounced development abnormalities.

The main method for analyzing the number and structure of chromosomes is karyotyping. This study involves several steps: cell culture; Preparation of preparations of metaphase chromosomes and interphase nuclei, colouring of the preparation; microscopic analysis. To detect numerical and structural changes in chromosomes, it is sufficient to carry out karyotyping using differential staining methods. In order to detect small deletions or duplications on the chromosome, it is generally necessary to use an in situ fluorescent hybridization technique that is sufficiently complex and cost-effective. economically costly

Among molecular genetic methods for analysis of chromosomal anomalies, the multiplex ligation-dependent probe amplification (MLPA) method is identified. MLPA is a variation of the multiplex polymerase chain reaction that permits amplification of multiple targets with only a single primer pair. It detects copy number changes at the molecular level, and software programs are used for analysis. Identification of deletions or duplications can indicate pathogenic mutations, thus MLPA is an important diagnostic tool used in clinical pathology laboratories worldwide. This technique allows to estimate the number of copies of the gene, detect point mutations, prolonged deletions or duplication of chromosomes. In a single assay, it is possible to determine the number of copies of up to 40 sites of different genes. It is not necessary to use living cells for analysis, which gives a time advantage and allows the detection of pathologies after long storage of the material.

The aim of the work was to determine the number of sexual and some somatic chromosomes and to analyze for common microdeletion syndrome in patients with various pathologies and a control group of people by MLPA method.

Commercial sets from MRC-Holland (Amsterdam, Netherlands) were used. The analysis was carried out using an automatic CEQ8800 capillary electrophoresis system (Beckman Coulter, USA).

The following chromosome structure disorders were identified: deletion 22q11.2 (Di Georgie syndrome), deletion 17p13.3 (Miller-Dicker syndrome), X chromosome dysomy in male XXY (Klinefelter syndrome). The use of molecular genetic methods for the diagnosis of hereditary diseases has a number of advantages over cytogenetic methods and allows the effective diagnosis of chromosomal diseases in both the prenatal and postnatal period.

Keywords: multiplex, ligation-dependent probe amplification, aneuploidy, microdeletion syndrome.

REFERENCES

THE FREQUENCY AND DISTRIBUTION OF SISTER CHROMATID EXCHANGES (SCES) IN THE INDIVIDUAL CHROMOSOMES OF HUMAN KARYOTYPE

Aytakin Hasanova, Gulnara Kuliyeva

1Azerbaijan Medical University, 2Baku Medical Plaza Clinic.

The aim of this study was to determine the frequency and distribution of sister chromatid exchanges (SCEs) in the individual chromosomes of human karyotype. Chromosomes were studied in (i) healthy subjects, (ii) subjects with rearrangements of X chromosome, (iii) in lymphoblastoid cell lines isolated from the peripheral blood of patients with acute leukaemias. 5-bromo-2-deoxyuridine (BUDR) was added for 48, 72 and 96 hours, respectively, in a concentration of 30 µg per ml. The slides were stained according to the technique of Perry and Wolff (4).

The average number of SCEs was 9.2 with no statistically significant differences between the individual groups. Out of the total number of SCEs 20% was found in the centromeric region with no difference between the cells in the 2nd and 3rd divisions. The observed distribution of breakpoints was approximately proportional to the relative length of individual chromosomes with a higher number in long chromosomes and a lower number in the small ones. Non-random distribution of SCEs was only found in the B group of chromosomes of lymphoblastoid cell lines, which showed an excess compared with the SCEs of both the controls and the expected frequency based on the relative length of chromosomes. Neither in the late replicating i(Xq) nor in the early replicating Xq— did the number of SCEs significantly exceed the expected value.

MATERIAL AND METHODS

The evaluation of the number and distribution of SCEs was performed in Seventy-five mitoses with karyotype 46,X,1 (Xq) derived from 4 patients. In one of these subjects a double centromere in the i(Xq) was identified by means of C banding. Twenty mitoses of one patient with reciprocal translocation 46, XX, t (X,4) (Xqter→Xq22: :4p16→4qter). Autoradiography proved that both the deleted X and the B4 chromosome with translocation were early replicating. Fifty-six mitoses obtained from three lymphoblastoid cell lines, derived from the peripheral blood of patients suffering from acute leukaemias. These were classified as dedifferentiated (Epstein-Barr virus positive), lymphatic and myeloid (both EBV-negative). One hundred and seventy mitoses from the control group consisting of 3 males with karyotype 46,XY and 4 females with 46,XX karyotype. The mean age of controls was 29.5 years. The peripheral blood leukocytes were cultivated for 48, 72 and 96 hours in EPL or Parker medium (Usol, Prague), enriched by 20% calf serum, with PHA (Welcome) and protected by streptomycin and penicillin. Bromodeoxyuridine (Sigma) was added in a concentration of 30 µg/ml since the beginning of cultivation. The cells were protected from light to avoid photolysis. Colcemid (Ciba) in a concentration of 10 µg/ml was added two hours before harvesting. The cells were hypotonized by 0.075 M KCl and methanol: acetic acid (3:1) were used for fixation(1).

The lymphoblastoid cell lines were grown as permanent suspension cultures from peripheral blood. They were established and subcultured in RPMI 1,640 medium enriched by 20% fetal calf serum, protected by streptomycin and penicillin, without PHA. BUdR was added in a concentration of 30 µg/ml for 48 or 72 hours following 48 hours of subcultivation. The examination of SCEs was carried out in the 15th, 3rd, and 38th, and in the 16th passages in three individual lines. The harvesting of chromosomes was the same as with the above-mentioned short-term peripheral blood cultures(2). Chromosomal preparations were stained according to the FPG staining technique of Perry and Wolff (4). Intact mitoses with harlequin chromosomes were photographed and SCEs evaluated first directly under the microscope, then from enlarged negatives or karyotypes. Exchanges occurring in short arms, long arms and in centromeric regions were counted separately. The number of SCEs was examined independently by two experienced observers and expressed as the number of breakpoints. Statistical evaluation was performed by means of the test and χ² test.

RESULTS

The difference in the number of SCEs found per cell in 50 mitoses of control subjects when evaluated from photomicrographs or karyotypes is small but significant (p<0.01). Therefore, the data described have been obtained from karyotypes only. The mean value of 9.5 SCEs per mitosis found in the control group was in no way significantly different from mean values of the other groups, i.e. those consisting of pathological karyotypes and lymphoblastoid cell lines (p>0.05).

High frequency of SCEs (20%) was found in the centromeric region of chromosomes after the 2nd and 3rd divisions in BUDR medium(6). The distribution of SCEs both observed and expected on the basis of the relative length of chromosomal groups in the karyotype with results of χ² test is shown in Table II. The changes were similar in all observed groups. The only exception

THE FIRST INTERNATIONAL SCIENTIFIC PRACTICAL VIRTUAL CONFERENCE HUMAN GENETICS AND GENETIC DISEASES: PROBLEMS AND DEVELOPMENT PERSPECTIVES
were B group chromosomes of the lymphoblastoid cell lines, where the number of breakpoints increased significantly (p<0.01). In this particular chromosomal group the breakpoints leading to exchanges were distributed proportionally along the whole length of all chromosomes. Significant difference was found not only as against the expected number of SCEs but also as against the control group. The increased number of breakpoints on the long chromosomes was naturally matched by their decrease in small chromosomes(5).

We preferred to evaluate the non-banded chromosomes (G-banding considerably interferes with the accuracy of SCE calculation), the regions of breakpoints are only roughly delineated. Even so it is clear that some regions are more often involved in SCEs than others. This is the case especially with regions 1q1, 3q2, 4q2, 8c and 16c. SCEs seem to be preferentially located on G-negative bands, as mentioned also by Morad, Jonasson and Lindsten (3).

REFERENCES

WORKING CONDITIONS AND HEALTH OF SEAFARERS

1Aytakin Hasanova, 2Simuzar Hajizada

1Azerbaijan Medical University. I Preventive Medicine Faculty. PhD in Medical Biology.


ABSTRACT

Relevance of a problem
Modern development of the fleet is inextricably connected with the solution of the problem of preservation and improvement of seafarers’ health, improvement of their working, living and rest conditions. The most important condition for the health of seafarers is the provision of optimal habitat on the vessel. At the same time, the vessel should be considered as an artificial ecological closed system, which provides the crew with a long-term active existence. A complex of interconnected environmental factors (climatic conditions of the navigation area, microclimate of ship 's rooms, noise, vibration, electrostatic radiation, electromagnetic radiation, harmful substances in the air, microflora in the room, psychophysiological factors, etc.) simultaneously acts on the human body in the conditions of buoyancy. The person in the final outcome responds to the environment as a whole. Therefore, the criterion reflecting the influence of the ship's environment on the human body is the level of functional state of the worker and his health [Lane T, et al., 2002; Rohrer JE. 2004; Heistaro S, et al., 2001; Jensen OC, et al., 2001; Thomas M, 2003].

There are currently poor working conditions and a high rate of occupational morbidity among maritime workers. The intensification of labor in the flight is increasing. There is poor quality of pre-trial and pre-trial medical examinations, reduction of medical positions on ships, leading to a deterioration of the quality of medical care or its complete absence. Obsolete vessels with expired service life are used. These circumstances lead to a deterioration of the habitat on ships and pose a threat to the health of workers. However, research on comprehensive hygienic assessment of working conditions and on the state of health of seafarers in modern conditions is scarce and often contradictory. This justifies the relevance of conducting a scientific study on the further study of the complex of factors that form the conditions of habitation on ships.

Work purpose
Develop a modern system of science-based measures to prevent adverse effects of ship environment factors on the human body to preserve the health and high efficiency of seafarers.

Research problems
To carry out a hygienic assessment of the physical and chemical factors forming the conditions of habitation on sea-going vessels, to assess the severity and intensity of work on ships of the sea-going fleet;
Undertake a comprehensive assessment of seafarers health;
To carry out an analysis of industrial injuries on ships of the marine fleet;
Provide scientific justification for the system of preventive measures and recommendations for the protection of labour and health of marine vessels in the modern period.

Methods of research
included hygienic, psychophysiological, sociological (questionnaire and interviewing) and analytical.

Provisions for protection
The leading adverse production factors on ships should be considered noise, vibration, microclimate parameters, lack of recovery, tension and severity of work;
Work in harmful conditions of production contributes to the development of changes in the state of health of seafarers, determines the structure of production - due to and occupational morbidity, increases the risk of occupational injuries;
The condition of labour and health of ship specialists justifies the need to organize a system of labour protection and health of seafarers, the main purpose of which is to create safe working conditions that exclude or minimize the risk of an employee receiving a professional disease or accident, preserving the life and health of ship specialists.

Scientific novelty of work
For the first time in the conditions of the Azerbaijani region, a comprehensive sanitary and hygienic assessment of working conditions, an analysis of the state of health and industrial injuries of specialists working on sea vessels was carried out.
Leading harmful production factors have been identified, and features of occupational and production-related morbidity have been identified.
For the first time, an assessment of the natural resistance of the body of seafarers was given, as well as an analysis of individual risk factors. On this basis, a system of labour protection and health of seafarers is justified and developed.

The practical value of the work lies in the fact that the comprehensive assessment of working conditions and the state of health of seafarers proposes a system of labour protection and health of persons working on sea vessels, which will ensure working conditions that meet the requirements of safety, which will contribute to the preservation of the life and health of maritime transport workers.

REFERENCES

2. Rohrer JE. Medical care usage and self-rated mental health. BMC Public Health 2004;4:3
FAMILY DOCTOR AND MEDICAL GENETICS

1Mustafa Salikhov, 2Kamile Salikhova

1Head of the Department of Family Medicine. Associate Professor.
2Assistant of the Department of Family Medicine.
1,2Azerbaijan Medical University

ABSTRACT
Introduction of the concept of family doctor as a primary medical care, involves his methods of work with a family as with a patient, the concept “family-patient”, introducing absolutely new state of things in organization and professional work of family doctor, as known, that structure of the family includes- size of the family, social composition and number of generations.
Now, it is very actual the medical insurance and preventive medicine in Azerbaijan, so importance of medical genetics is increasing. The family doctor should do clinical and geneological investigations to the patient and to the members of his family, find out the group of the patients with the high risk and involve the preventative measures.
Keywords: family doctor, preventive medicine, clinical and geneological investigations.

Clinical genetics is one of the chapters of clinical medicine and human genetics. Clinical genetics solving problem of diagnosis, prognosis and treatment of different hereditary diseases.
Introduction of the concept of family doctor as a primary medical care, involves his methods of work with a family as with a patient, the concept “family-patient”, introducing absolutely new state of things in organization and professional work of family doctor, as known, that structure of the family includes- size of the family, social composition and number of generations.
Now, it is very actual the medical insurance and preventive medicine in Azerbaijan, so importance of medical genetics is increasing.
The family doctor should do clinical and geneological investigations to the patient and to the members of his family, find out the group of the patients with the high risk and involve the preventative measures.
Therefore, knowledge of medical and clinical genetics – one of the necessary condition in professional activity of a family doctor.
Family doctor must have certain skills in taking patient's anamnesis, knowledge of signs in hereditary pathology and should inform families of inappropriateness of close blood related marriages, because children born from close blood related marriages have more possibilities of in hereting genetic diseases and birth defects and newborn mortality is more higher.
Certainly, invasion of family doctor to the area of the patient’s hereditary structure possible only with one aim – to treat the patient and to create condition, when the birth defect gen wouldn’t transfer to the children. And finally, theoretical calculations of family geneticist and experience in genetical consultations give an opinion regarding rise of having a sick baby in every particular family and find people, who need medical genetic consultation. Important part of care in patients with genetic disease, in addition to therapeutic and preventative measures, are also psychotherapy correction and social rehabilitation.
Based on above, it is important to note: qualification of the doctor and his participation in the life of the family affect not only the health of people who live now, but also the health and prosperity of future generations.

REFERENCES
1. Integrating Genetic Counseling into Family Medicine JEFFREY R. MARTIN, M.D. ADAM S. WILIKOFSKY, PH.D.Lancaster General Hospital Family Medicine Residency Program Lancaster, Pennsylvania Am Fam Physician. 2005
2. Genetics for Family Physicians W. Gregory Feero, MD, PhD
THE IMPORTANCE OF ELECTRON MICROSCOPIC SYSTEMATIC CLASSIFICATION WE OFFER IN CERVICAL CANCER

Aynur Panahova

Department of Oncology, Azerbaijan Medical University.

ABSTRACT

Based on the electron microscopic diversity in tumour cells in cervical cancer, we systematized the results obtained in our study, considering the difference in the various cells of biopsies taken from the same patient. The scientific novelty of the work is the development of an ultrastructural microscopic classification of neoplastic cells in cervical cancer by summarizing electron microscopic signs. The said classification allows us to explain the electron microscopic theoretical basis of differential diagnosis based on the organ-tissue and cytospecific ultrastructural signs (groups and types of tumour cells) of tumour tissue, on the one hand. On the other hand, it helps to specify the degree of ultrastructural differentiation of neoplasms, comparison between cells, types of tumour cell and options of tumours. The main purpose of the use of electron microscope in practical medicine is related to the histogenetic diagnosis of tumours. From this point of view, the systematic classification we offer allows us to systematize electron microscopic signs of tumour cells and tumour cell groups, on the one hand, and, on the other hand, it allows us to detect the degree of ultrastructural differentiation of new derivatives (tumour cells, tumour cell groups, ratio of tumour options). Thus, the systematic classification we provided as a result of our study allows us to explain the theoretical basis of the general principles of tumour cells. This system explains the specialization of ultrastructural organ-tissue-cell during specific electron microscopic differentiation of tumour cells. We can also get information, at electron microscopic level, on the morphogenetic potential of cambial cells, which are the source for creation of tumour cells as a result of such a systematic analysis.

Thus, in the irregular and numerous ultrastructural options of tumour cells in the tumour mass, precise regularities can be obtained, and we can say something about their cell composition based on these regularities. Based on the stated and obtained certain results, it is possible to substantiate the great importance of electron microscopy in practical activity and theoretic studies, diagnostics, differential diagnostics of tumours, differentiation, histogenesis, biological features of neoplastic cells.

OBJECTIVE

The research aims to optimize the prognostic criteria of the disease based on a comprehensive clinical, instrumental, morpho-functional, and statistic analysis of cervical cancer, as well as electron microscopic indicators.

MATERIAL AND METHODS

The study covered 330 patients with in cervical background diseases, precancerous diseases and cancer from 2007 to 2017.

Main group: 220 patients with cervical cancer;
Control group: 110 patients - erosions, cervical polyps, CIN 1-3

Electron microscopic studies were carried out and based on these studies, cytological (Pap-Smear and Bethesda) and histological and electron microscopic study of the ultrasound classification of Cervical Neoplasia was carried out. Electron microscopic studies were conducted on 93 cancer patients in an electron microscopic laboratory at the Department of Histology of AMU.

RESULTS

Differentiated tumour cells rich (60%) with ultrastructural organo-tissue and organoids with cytospecific signs are predominant in cervical squamous cell corned cancer (option 1) [6, 62, 117]. The amount of organoids in cervical squamous cell non-corned cancer is at medium level (50%), the activity of 2 groups of non-differentiated tumour cells, ultrastructural cataplasias noticed in one or more types of cell indicate a worse prognosis (option 2) [6, 62, 117]. In non-differentiated cervical cancer, only non-differentiated tumour cells with less ultrastructural organoids (30%) were noticed (option 3) [6, 62, 117]. The same number of differentiated and non-differentiated tumor cells is detected in cervical adenocarcinomas (variant IV). The said elements are related to heterogeneous cells and contribute to progress, development of tumours, and metastasis [6, 62, 117].
Based on the ultrastructural classification we applied in cervical neoplasms, different types, species of tumour cells, and comparative tumour options in the groups to which they belong were identified [2, 3, 117]. Based on calculation of prognostic score, it was detected that those over the age of 45 years (informativeness - 0.821) had more abortions in their anamnesis (1,640) had HPV positive (1,145), high stage (2,529), low degree of differentiation (1,422), lymph nodes (4,773) and gave distant metastases (4,823), and patients with less organelles (2,404) were found to be more likely to have recurrence during electron microscopic examination [3].

REFERENCES

ASSOCIATION OF RS7903146 C/T POLYMORPHISM OF TCF7L2 GENE WITH TYPE 2 DIABETES MELLITUS IN AZERBAIJAN POPULATION: PRELIMINARY STUDIES

Zarintaj Rustamova, Lala Akhundova, Gulmira Alibayova, Nurmammad Mustafayev, Irada Huseynova

Institute of Molecular Biology & Biotechnologies, Azerbaijan National Academy of Sciences.

AIM
Nowadays, type 2 diabetes mellitus (T2DM) is one of the most propagative endocrine diseases. Prediagnosis of T2DM, which is developing due to the combination of genetic and lifestyle factors, is an urgent problem of the diabetology. The aim of this work was to study the association of the single nucleotide polymorphism (SNP) rs7903146 C/T of TCF7L2 gene with a risk of developing of the T2DM.

MATERIALS AND METHODS
The object of the study were DNA samples, isolated from patients with T2DM and healthy individuals (approximately equal amount in each group of ~100). DNA profiles were obtained by PCR using primer pairs (marker rs7903146): forward 1 (C allele): 5′-GAACAATTAGAGCTAAG CACTTTTTAGAAAC-3′; forward 2 (T allele): 5′-GAACAATTAGAGCTAAGCAGCCTTTTT AGAGAT-3′ and common reverse: 5′-AGATGAG ATGTAGCAGTGAAGTGC-3′. PCR conditions were determined by gradient PCR.

RESULTS
The results of the current study revealed the following gene variants (in %): homozygotes CC 22.7 and 40.9, homozygotes TT 45.5 and 22.7, heterozygotes CT 31.8 and 36.4 in experimental and control groups respectively. Allele frequencies: \(C_{\text{exp}}=0.386, \; T_{\text{exp}}=0.614; \; C_{\text{control}}=0.591, \; T_{\text{control}}=0.409.\) The values of the ratios \(C_{\text{exp}}:T_{\text{exp}}=0.63; \; C_{\text{control}}:T_{\text{control}}=1.44; \; C_{\text{exp}}:C_{\text{control}}=0.65; \; T_{\text{exp}}:T_{\text{control}}=1.5\) indicate that the presence of the C allele in the control and the T allele in the experimental group is ~1.5 times greater than that in experimental and control group respectively. In addition, TT homozygotes in the experimental group are 2 times more than in control group. The number of heterozygotes (CT) is almost equally.

CONCLUSIONS
Despite that the frequency of the T allele and the number of TT homozygotes in the group with T2DM are noticeably higher than that in the control, statistically significant correlation between rs7903146 polymorphism and T2DM was lower than that was expected (\(p<0.1\)). Since T2DM is a polygenic disease, this may be because of the sample size, which was not enough to make a clear conclusion about the association. Moreover, studies of additional markers should be included.

Keywords: molecular-genetic markers, SNP polymorphism, type 2 diabetes mellitus (T2DM), gene transcription factor 7 like 2 (TCF7L2), homozygote, heterozygote.

REFERENCE
MOLECULAR GENETIC ANALYSIS OF GLA GENE CAUSING INHERITED FABRY DISEASE FOR POPULATION OF AZERBAIJAN REPUBLIC

Narmin Salayeva

Doctoral student of the Department of Biology and Basics of Medical Knowledge.

Lankaran State University, Lankaran, Azerbaijan.

ABSTRACT

For the first time in Lenkoran-Astara administrative area of Azerbaijan Republic, patients with cardiomiopathies were genetically screened for Fabry metabolic disease. Screening was carried out by means of identification of α-galactosidase enzyme activity and quantity of globotriasylphosphogosine.

In 12 out of 29 examined persons we got activity deficit of α-galactosidase enzyme, and amount of globotriasylphosphogosine was higher than the norm which were specific for Fabry disease. In 8 women manifested X-linked inheritance type as heterozygotes for Fabry disease, and 4 men were identified as hemizygoes.

Molecular genetic analysis identified two different mutations of GLA gene: 801+3A>G mutation in intron 5 and, substitution of Adenine nucleotide with Guanine nucleotide in position 137 (137,A>G) of GLA gene. To prevent Fabry disease it is recommended to screen affected persons’ family members for α-galactosidase enzyme activities.

Keywords: Fabry, inherited disease, mutation, α-galactosidase, lisosome, globotriasylphosphogosine (lyso-Gb3), GLA

INTRODUCTION

Fabry disease (Anderson-Fabry disease, inherited distonic lipoidosis etc.) being a lisosome metabolic disease relates to a group of orphan (rare) diseases[1]. GLA gene, modifying Fabry disease clinic, locates on the X-sex chromosome long shoulder - Xq22. GLA gene encodes α-galactosidase enzyme. Up to nowadays more than 640 mutations were identified. Mutations 70% are related to missense or nonsense mutations group. The most of mutations have family specificity[5]. Fabry disease being one out of 60 diseases of lisosome storage diseases is a result of complete or partial α-galactosidase A enzyme activity deficiency. Alpha-galactosidase A enzyme activity reduction causing sphingolipidoses metabolism damage, leads to storage of globotriasylphosphogosines. The disease has broad spectrum of simptoms. In the course of disease one organ is mainly damaged, problems are in heart or kidney [4].

It should be mentioned that because population of Azerbaijan Republic have never been diagnosed for Fabry disease, the disease has never been identified, there is no data on clinic, biochemistry and genetics of the disease.

The goal of the article was to screen patients with cardiomiopathies for Fabry inherited lisosome metabolic disease and to carry out molecular genetic analysis of GLA gene for identified patients.

MATERIAL AND METHODS

29 patient suspicious for Fabry inherited lisosome disease venous blood samples 1ml each were taken into EDTA anticoagulant sample tube and then absorbed into special DBS (dry blood spots) cards. Cards with absorbed blood samples stay at room temperature for 1 hour, and then are analysed at the Chair of Laboratory Science (Azerbaijan State Doctors’ Advanced Training Institute after A.Aliyev) and CENTOGENE laboratories (Germany, Rostock city).

29 cardiologic patients from Lenkoran, Masalli and Astara Central Regional Hospitals were suspicious for Fabry disease to develop those cardiomiopathies. For that purpose α-galactosidase enzyme activities were measured, and in case of enzymatic deficit amounts of globotriasylphosphogosine (lyso-Gb3) were identified[2]. Fluorimetric method and liquid chromatography were used in genetic screening, Sanger methodique was applied for direct sequencing of GLA gene. Testing of the existing mutation in GLA gene became possible with this method. The method was developed in CENTOGENE laboratories, Rostock, Germany[5].

RESULTS

In Masalli region 4 patients identified were 2 men and 2 women. In brothers T.I. and T.A. α-galactosidase enzyme activity showed lower than the norm activity as 0.8 mkmol/l/s (N ≥15.3 mkmol/l/s). To verify the diagnostics, another test was carried out for lyso-Gb3 amount, and happened to be higher than the norm - 106.0 ng/ml (T.I.) and 106.0 ng/ml (T.A.). Fabry disease having X-linked chromosome dominant inheritance type, both of brothers were hemizygoes carriers of GLA gene. Alpha-galactosidase enzyme deficiency for sisters T.G. and A.G. being 3.2 mkmol/l/s, 1.9 mkmol/l/s, relatively, amounts of Lyso-Gb3 for them were vice versa higher – 15.6 ng/ml (T.G) and 8.3 ng/ml (A.G). For both of sisters X-linked disease one organ is mainly damaged, problems are in heart or kidney.

In 12 out of 29 examined persons we got activity deficit of α-galactosidase enzyme, and amount of globotriasylphosphogosine was higher than the norm which were specific for Fabry disease. In 8 women manifested X-linked inheritance type as heterozygotes for Fabry disease, and 4 men were identified as hemizygoes.

Molecular genetic analysis identified two different mutations of GLA gene: 801+3A>G mutation in intron 5 and, substitution of Adenine nucleotide with Guanine nucleotide in position 137 (137,A>G) of GLA gene. To prevent Fabry disease it is recommended to screen affected persons’ family members for α-galactosidase enzyme activities.

Keywords: Fabry, inherited disease, mutation, α-galactosidase, lisosome, globotriasylphosphogosine (lyso-Gb3), GLA

INTRODUCTION

Fabry disease (Anderson-Fabry disease, inherited distonic lipoidosis etc.) being a lisosome metabolic disease relates to a group of orphan (rare) diseases[1]. GLA gene, modifying Fabry disease clinic, locates on the X-sex chromosome long shoulder - Xq22. GLA gene encodes α-galactosidase enzyme. Up to nowadays more than 640 mutations were identified. Mutations 70% are related to missense or nonsense mutations group. The most of mutations have family specificity[5]. Fabry disease being one out of 60 diseases of lisosome storage diseases is a result of complete or partial α-galactosidase A enzyme activity deficiency. Alpha-galactosidase A enzyme activity reduction causing sphingolipidoses metabolism damage, leads to storage of globotriasylphosphogosines. The disease has broad spectrum of simptoms. In the course of disease one organ is mainly damaged, problems are in heart or kidney [4].

It should be mentioned that because population of Azerbaijan Republic have never been diagnosed for Fabry disease, the disease has never been identified, there is no data on clinic, biochemistry and genetics of the disease.

The goal of the article was to screen patients with cardiomiopathies for Fabry inherited lisosome metabolic disease and to carry out molecular genetic analysis of GLA gene for identified patients.

MATERIAL AND METHODS

29 patient suspicious for Fabry inherited lisosome disease venous blood samples 1ml each were taken into EDTA anticoagulant sample tube and then absorbed into special DBS (dry blood spots) cards. Cards with absorbed blood samples stay at room temperature for 1 hour, and then are analysed at the Chair of Laboratory Science (Azerbaijan State Doctors’ Advanced Training Institute after A.Aliyev) and CENTOGENE laboratories (Germany, Rostock city).

29 cardiologic patients from Lenkoran, Masalli and Astara Central Regional Hospitals were suspicious for Fabry disease to develop those cardiomiopathies. For that purpose α-galactosidase enzyme activities were measured, and in case of enzymatic deficit amounts of globotriasylphosphogosine (lyso-Gb3) were identified[2]. Fluorimetric method and liquid chromatography were used in genetic screening, Sanger methodique was applied for direct sequencing of GLA gene. Testing of the existing mutation in GLA gene became possible with this method. The method was developed in CENTOGENE laboratories, Rostock, Germany[5].

RESULTS

In Masalli region 4 patients identified were 2 men and 2 women. In brothers T.I. and T.A. α-galactosidase enzyme activity showed lower than the norm activity as 0.8 mkmol/l/s (N ≥15.3 mkmol/l/s). To verify the diagnostics, another test was carried out for lyso-Gb3 amount, and happened to be higher than the norm - 106.0 ng/ml (T.I.) and 106.0 ng/ml (T.A.). Fabry disease having X-linked chromosome dominant inheritance type, both of brothers were hemizygous carriers of GLA gene. Alpha-galactosidase enzyme deficiency for sisters T.G. and A.G. being 3.2 mkmol/l/s, 1.9 mkmol/l/s, relatively, amounts of Lyso-Gb3 for them were vice versa higher – 15.6 ng/ml (T.G) and 8.3 ng/ml (A.G). For both of sisters X-linked disease one organ is mainly damaged, problems are in heart or kidney.

In Masalli region 4 patients identified were 2 men and 2 women. In brothers T.I. and T.A. α-galactosidase enzyme activity showed lower than the norm activity as 0.8 mkmol/l/s (N ≥15.3 mkmol/l/s). To verify the diagnostics, another test was carried out for lyso-Gb3 amount, and happened to be higher than the norm - 106.0 ng/ml (T.I.) and 106.0 ng/ml (T.A.). Fabry disease having X-linked chromosome dominant inheritance type, both of brothers were hemizygous carriers of GLA gene. Alpha-galactosidase enzyme deficiency for sisters T.G. and A.G. being 3.2 mkmol/l/s, 1.9 mkmol/l/s, relatively, amounts of Lyso-Gb3 for them were vice versa higher – 15.6 ng/ml (T.G) and 8.3 ng/ml (A.G). For both of sisters X-linked disease one organ is mainly damaged, problems are in heart or kidney.

In Masalli region 4 patients identified were 2 men and 2 women. In brothers T.I. and T.A. α-galactosidase enzyme activity showed lower than the norm activity as 0.8 mkmol/l/s (N ≥15.3 mkmol/l/s). To verify the diagnostics, another test was carried out for lyso-Gb3 amount, and happened to be higher than the norm - 106.0 ng/ml (T.I.) and 106.0 ng/ml (T.A.). Fabry disease having X-linked chromosome dominant inheritance type, both of brothers were hemizygous carriers of GLA gene. Alpha-galactosidase enzyme deficiency for sisters T.G. and A.G. being 3.2 mkmol/l/s, 1.9 mkmol/l/s, relatively, amounts of Lyso-Gb3 for them were vice versa higher – 15.6 ng/ml (T.G) and 8.3 ng/ml (A.G). For both of sisters X-linked disease one organ is mainly damaged, problems are in heart or kidney.
autosomal dominant inheritance type in heterozygous state was identified. Patient M.A. originated from Lerkoran had got alpha-galactosidase activity lower than the norm – 2.3 mkmol/l/s, but amount of Lyso-Gb3 was higher than the norm (109 ng/ml).

In Astara area in the family of 6 persons were women and one man born on 12.06.1978. Alpha-galactosidase enzyme showed "0" activity for patient K.A., and amount of Lyso-Gb3 was much higher than the norm (218.0 ng/ml). In six women of the family, activity of alpha-galactosidase enzyme was in range of 1.4-1.8 mkmol/l/s. The mean amount of Lyso-Gb3 was registered as 16.0 ng/ml (11.0-21.0 ng/ml).

Masalli area all patients were members of one and the same family, thus they had got the same mutation 801+3A>G of GLA gene.

In patient M.A. molecular genetic diagnostics of GLA gene identified substitution of Adenine nucleotide with Guanine nucleotide in position 137 (137, A>G). This mutation in position 137 of GLA gene at the time of Adenine nucleotide substitution with Guanine nucleotide causes substitution of Histidine amino acid with Arginine amino acid in position 46 in the course of protein (enzyme) biosynthesis (46 His>Arg).

One and the same GLA gene mutation was identified in one family in Astara area. Adenine nucleotide was substituted with Guanine nucleotide in position 137(137,A>G) of GLA gene. Mutation happened in the gene caused the change of Histidine amino acid to Arginine amino acid (46 His>Arg) in position 46 while protein biosynthesising. Because of mutation relates to missense (nonsense) mutation type, the synthesis of enzyme was completely destroyed.

So, twelve patients with Fabry inherited metabolic disease were identified resulting from genetic screening in Astara, Lenkoran and Masalli areas population especially among cardiomiopathic patients for α-galactosidase enzyme and globotriasylsphingosine. Molecular genetic analysis of GLA gene identified two different mutations: 801+3A>G and 137, A>G. Both mutations of GLA gene were found for the first time for Republic population. To prevent Fabry disease, index patients family members are recommended to pass through screening of α-galactosidase enzyme activity.

REFERENCES

A. Волгина С. Я. Болезнь Фабри // Практическая медицина (научно-практический медицинский журнал), 2012, 62, 7, 75-79.
HUMAN AS A BIOTIC FACTOR IN ACCUMULATION OF MICROELEMENTS WITH WEAK TOXICITY IN BONE AND MUSCLE TISSUE OF BACKGROUND REPTILE SPECIES IN THE ABSHERON PENINSULA

Aysel Hashimova
Azerbaijan Medical University, The Department of Medical Biology and Genetics. Azerbaijan.

ABSTRACT

The accumulation and amount of microelements with weak toxic effects have been studied in bone and muscle tissue of following Reptile species - Water snake (Natrix tessellata Laurenti, 1768), Mediterranean turtle (Testudo graeca L.1758) and Caspian bent-toed gecko (C.caspius E. 1831). Studied microelements are cobalt Co, molybdenum Mo, manganese Mn, and chromium Cr. All the four microelements that we have studied are mostly accumulated in the body of Caspian bent-toed gecko. It should be noted that the Caspian bent toed gecko is very small and more functionally active among background reptile species that we studied. Its occurrence in oil, gas wells and in areas exposed to technogenic and antropogenic pollution, indicates its plasticity and resistance to toxic microelements.

INTRODUCTION

In recent years the Absheron peninsula has been subjected to anthropogenic and technogenic pollution in addition to the intense urbanization process.[5] So, it affects all components of the biocenosis in the peninsula and especially, to the flora and fauna. The reptiles that have historically been settled here cannot stay away from these effects, as they feed on existing plants, native invertebrates and vertebrates and drink the water of this area. Therefore, their bodies are rich in microelements of the peninsula, respectively. The problem of deficiency and excess of microelements in human and animal organism is the important issue of world scientists. Thus in both cases, pathological processes occur. The reason for accumulation of toxic microelements in the body of research objects is anthropogenic and technogenic pollution and its directly related to human activity. In this research we aimed to determine how human impacts on the research objects and which of these objects is mostly exposed to this impact.

MATERIALS AND METHODS

The objects of our research are the Mediterranean turtle (Testudo graeca L.1758) from the order Turtles (Testudines), the water snake (Natrix tessellata Laurenti, 1768) from the order Snakes (Serpentes) and the Caspian bent-toed gecko (C.caspius E. 1831), from the order Lizards (Sauria). All these orders belong to the class of Reptiles. The route method was used for the collection of materials. The amount of the following microelements -cobalt, molybdenum, chromium, and manganese, which have weak toxic effects and are needed for the normal functioning of the body, have been studied in the muscle and bone tissue of the selected reptile species. [1,2,6] These microelements have toxic effects when their amount is higher than the norm, and in the case of their deficiency, the functional activities of the body are impaired. However, the proper amount of these microelements is actively involved in regulating the physiological and biochemical functions of the body.

For the biochemical analysis 10 specimens were taken from each of the Caspian bent-toed geckoes and water snakes, as well as 7 specimens from the Mediterranean turtles. It should be noted that, taking into consideration the sharp decline in the number of Mediterranean turtles in recent years and their inclusion in International Union for Conservation of Nature’s Red List of Threatened Species, samples of the turtles have been taken from bone and muscle tissue of damaged or dead bodies that were found on the roadside. Furthermore, during the extraction of the material from the Caspian bent-toed gecko, it was sampled from both muscle and bone tissues for the chemical analysis of microelements, because, due to its small quantity it was not well anatomized. Accordingly, the muscle and bone tissue of the water snakes that we investigated were analyzed together. Since both tissues are a major component of the supporting apparatus, it is more advisable to perform this analysis by using both muscle and bone tissue. Bone tissue and somatic muscles (that form a separate tissue group) are important for the body due to their biological and physiological properties. Considering this point, we aimed to study the amount of microelements with weak toxicity that have been found in both tissues.

The quantitative analysis of the collected material was carried out at the Institute of Radiation Problems of ANAS in the Laboratory of “Physics and Chemistry of Harmful Impact on the Environment”. The analysis was performed using the AAS-Atomic Absorption Spectroscopy method. Atomic Absorption Spectrometer 220 FS was used for determining the accumulation and quantity of toxic microelements in the bone and muscle tissue of the animals we investigated. The advantage of Atomic Absorption Spectrometer is that it is possible to identify several elements in the same solution with high sensitivity, selectivity, and spending little time.
RESULTS

Cobalt is one of the most important micronutrients for the vital functions of the body. Interest in this micronutrient started in the 30s of the last century. Large horned animals and sheep in various countries around the world, including Russia, Canada, New Zealand, Scotland and Australia, have been suffered from severe diseases such as weight loss, loss of appetite, fatigue and anemia, and sometimes it resulted in death. Scientists initially attributed the occurrence of these symptoms to the lack of iron in the diet. In the late twentieth century, it was discovered that the addition of small amounts of cobalt to the diet would help to cure all the above-mentioned diseases. Cobalt micronutrients have been included in the list of cancerous factors by the International Health Organization because of their involvement in the development of malignant tumors.[7]

Among the reptiles we investigate, cobalt microelement is more commonly found in muscle and bone tissue of the Caspian bent-toed gecko (1,240 mg / kg). In the second place are water snakes (0.139 mg / kg), and in the third place are turtles (0.076 mg / kg). As it seems, the large amount of cobalt in bone and muscle tissue of the Caspian bent-toed gecko is due to its organism’s highly adaptive ability to these microelement. On the other hand, as the cobalt microelement actively takes part in the synthesis of muscle proteins and geckoes are more active among reptiles we investigated, it is logical that the cobalt mostly accumulated in the muscles of geckoes.

Manganese is one of the most important microelements for the organism, it is widely distributed in the air, water, soil, plants and animals.[3] It actively participates in many biological processes during the normal life and activity of both plants and animals. Manganese stimulates growth and development of the body, activates many enzymes, participates in the respiratory process, as well as in the metabolism of minerals. People should have 0.4-10 mg of manganese daily with food. Manganese microelement is highly concentrated in the bone and muscle tissue of the reptiles we investigate. Thus, the amount of manganese in the muscle and bone tissue of the Caspian bent-toed geckoes is 147.09 mg / kg, in water snakes 29.850 mg / kg and in Mediterranean turtles was 8.529 mg / kg. Comparison of the animals studied, it can be concluded that the high concentration of the manganese in the muscle and bone tissue of Caspian bent-toed gecko and water snake is depending on their daily nutrition, and on the other hand, both species are mostly associated with water (especially snakes).

The normal amount of chromium microelement has a high value for the organism.[4,6] The essential role of chromium in the body is to take part in synthesis of fats, to play an important role in the metabolism of sugar and its stability in the blood. The compounds of chromium with metals have no toxic effect, but its compounds in the form of solution are very toxic. The leading position for the amount of chromium in the muscle and bone tissue of the reptiles we investigate is the Caspian bent-toed gecko - 14.912 mg / kg. In the second place, the Mediterranean turtle - 4.461 mg / kg, and the last - the water snake - 0.965 mg / kg. The high concentration of chromium microelement in geckoes and turtles is consistent with the literature, however the reason of small amount of chromium in the snakes can be related to their nutrition with fish and other marine animals that contain small amount of chromium.

Molybdenum microelement is actively takes part in the activation of enzymes involved in detoxication of foreign substances.[3,7] It also regulates iron metabolism in the liver, and low doses stimulate the formation of hemoglobin, but large amounts of this microelement prevent hemoglobin formation. Molybdenum is most commonly found in the organs involved in the metabolism of the liver, kidneys, lymph nodes, and less concentrated in the muscle tissue. Another interesting feature of this microelement is that it, as an antagonist of copper, is involved in the exclusion of the copper from the liver and as an antagonist of phosphorus it takes part in exclusion of phosphorus from the bone tissue. The demand for molybdenum among animals varies depending on the species that it belongs to, its age and the amount of copper, zinc, lead and inorganic compounds in the food it receives. Usually the young organism is more susceptible to molybdenum than the elder one.

Quantitative analysis of the molybdenum microelement in muscle and bone tissue of the reptiles collected from the urbanized areas of the Absheron peninsula has resulted in the following findings: 0.557 mg / kg in Caspian bent-toed geckoes, 0.224 mg / kg in Mediterranean turtles and 0.254 mg / kg in water snakes. When comparing the amount of micronutrients with low toxicity in bone and muscle tissue, it is becoming clear that the molybdenum micronutrient is less common in the animals studied.

CONCLUSION

As mentioned above, the amount of microelements, that we studied in bone and muscle tissue of reptiles differs depending on species and microelements. Some of them are below, while others are above the standard average and it is related to disturbance of the ecological balance. First of all, these microelements are excessive in external environment and on the other hand, the amount of some microelements exceeds the norm in muscle and bone tissue of reptiles. So, it helps us to come to conclusion that these organisms have a high ability to accumulate microelements and adapt to them. All the four microelements that we have studied are mostly accumulated in the body of Caspian bent-toed gecko. It should be noted that the Caspian bent toed gecko is very small and more functionally active among background reptile species that we
studied. Therefore, high levels of microelements detected in the Caspian bent-toed gecko do not cause lethal effects. Toxic microelements in bone and muscle tissue of studied reptiles are higher than the standard average and one of the reasons why they do not have a lethal effect is that, these micronutrients are accumulated in functionally active bone and muscle tissue.

REFERENCES

3. Ivanova TM //Biological role and toxic effects of manganese// 2010, p-115-120
6. Kist AA //Phenomenology of biochemistry and bioorganic chemistry// Tashkent, -1987, p-236
7. Kovalenkov YK //The impact of chelates of cobalt, zinc, copper and iron on laboratory animals and large horned tur// The news of TAA-2011, vol.-1, p.-139-149
IMMUNOLOGICAL AND GENETIC ASPECTS OF HEREDITARY ANTIBODY DEFICIENCIES

1 Gulnara Nasrullayeva, 2 Vafa Mammadova, 3 Gunay Aliyeva, 4 Elnura Atakishiyeva

1 Head of Scientific-Research Immunology Laboratory. Professor.
2 Leading specialist of the Scientific-Research Immunology Laboratory. PhD.
3 Leading specialist of the Scientific-Research Immunology Laboratory.
4 Leading specialist of the Scientific-Research Immunology Laboratory.

ABSTRACT

INTRODUCTION AND AIM

Primary Immunodeficiencies (PID) are rare but very severe hereditary pathologies associated with disfunction of immune system. The most common type of PID is Antibody Deficiencies (AD) that characterized by the recurrent bacterial infections in gastrointestinal, respiratory and urinary tract.

The aim of the study was to analyze the data of pediatric and adult patients diagnosed as AD in our PID center between 2010-2019.

METHODS

All patients with PID were examined by blood chemistry and phenotyping of immune cells in peripheral blood, measurement of IgM, IgG, IgA, IgE and sIgA levels in serum and saliva, phagocytic activity by NBT, and detection of CIC by photometric method. Additional X-ray and ultrasound examinations were also performed.

RESULT

In 2010-2020 years in Research Immunology laboratory of Azerbaijan Medical University patients suspected to immune disorders were examined and the Antibody deficiency was detected in 46 patients: 3 adult and 6 children in age of 2-8 years were diagnosed with sIgA deficiency, Hyper IgM syndrome was rare deficiency-only in 2 children in age 1-2 years.

The biggest group of patients with common variable immune deficiency and agammaglobulinemia included children of different ages and adults. All the patients who had frequent and severe infections-lung, skin, gastrointestinal diseases, were receiving replacement therapy-IVIG. Immune disbalance show decrease of quantity and function of B-cell, hypogammaglobulinemia. Detailed diagnosis in 5 patients based on the genetic tests which were carried out. The following results show genetic mutations: 2 case – Btk deficiency, 2 cases - CD40 LG deficiency, 1 case - BLNK gene mutation.

CONCLUSION

The children with repeated, severe bacterial infections and hypoglobulinemia have to be suspected on PID especially to AD type. Immune examination should be including serum immunoglobulin levels and B and T-cell subsets in peripheral blood as the first stage of detection.

REFERENCE

EARLY EPILEPTIC ENCEPHALOPATHY GENETICS OF PATIENT IN AZERBAIJAN

Ziba Nasibova

Dr. Laboratory of Human Genetics. ANAS Genetic Resources Institute. Azerbaijan.

ABSTRACT

Blood genetic analysis of the patient M.D. identified mutation existence in four genes. M.D. The following synonyms are for ATP7A.
The ATP7A variant c.3632G>A p. (Arg1211Gln) causes an amino acid change from Arg to Gln at position 1211. Its classified as variant of uncertain significance (class 3) according to the recommendations of Centogene and ACMG (please, see additional information below).
The second gene - The FGF23 variant c.61G>A p. (Val21Ile) causes an amino acid change from Val to Ile at position 21. It is classified as variant of uncertain significance (class 3) according to the recommendations of Centogene and ACMG (please, see additional information below).
The third gene - The CTSA gene provides instructions for making a protein called cathepsin A. Cathepsin A can act as a protease, cutting apart other proteins in order to break them down. Cathepsin A can also act as a protective protein, interacting with other enzymes to prevent them from breaking down prematurely. Based on this protective function, this enzyme is also called protective protein/cathepsin A or PPCA.
The fourth gene - The PKLR gene is active (expressed) in the liver and in red blood cells, where it provides instructions for producing an enzyme called pyruvate kinase. This enzyme is involved in a critical energy-producing process known as glycolysis. During glycolysis, the simple sugar glucose is broken down to produce energy. Specifically, pyruvate kinase is involved in the last step of the glycolytic pathway. In this step, a cluster of oxygen and phosphorus atoms (a phosphate group) is moved from a molecule called phosphoenolpyruvate to another molecule called adenosine diphosphate (ADP), resulting in molecules called pyruvate and adenosine triphosphate (ATP). ATP is the cell's main energy source. Thus, apparently, combination of gene mutations as ATP7A, PKLR, CTSA, FGF23, in the child lead to disease – early epileptic encephalopathy.
Keywords: genetics, molecular genetic analysis, epileptic encephalopathies, ATP7A gene, FGF23, CTSA gene, PKLR gene.

INTRODUCTION

Epileptic encephalopathies (EE) are a group of progressing diseases of different etiology which is expressed as neurocognitive deficit and epileptiform activity on the electroencephalogram. EE make 15% of all epilepsy forms in childhood age and up to 40% of all epileptic onsets in their first 3 years of life. 10 syndromic forms of EE are outlined [5]. Genetic factors play special role in pathologies development in around 70—80% patients, and not less than 40% of all idiopathic epilepsies have got monogenic nature. 35 genes responsible for EE occurrence are identified, and the search is still continued. Severe genetic heterogeneity of early EE is showed, 16 of which are inherited autosomal-dominant, 13 – autosomal recessive, 4 – X-linked recessive and 2 – X-linked dominant. Differential approaches to some EE syndromes cure are presented [7,10]. Totally for nowadays the OMIM catalogue includes more than 400 genes, where mutations lead to occurrence of monogenic diseases with following seizures. Except that seizure syndrome is included in symptomatic complex of significant number of chromosomal syndromes diagnosed by means of standard karyotype as well as chromosomal micro matrix analysis [1-4].
Goal of our researches is modern molecular genetic diagnostics study of one patient with epileptic encephalopathy diagnosis from Azerbaijani family.

MATERIAL AND METHODS

Patient M.D. is a girl of 9 months with diagnosis of epileptic encephalopathy who was born from consanguine marriage where parents are first cousin sibs (their fathers are brothers). Patient M.D. is the third child and has got two sound elder brothers (17 and 19 years of age).
Blood sampling for genetic diagnostics was done on DBS (Dry Blood Spots) cards. After drying up blood spots, the sample was sent to CENTOGENE laboratories (Germany) for molecular genetic analysis.
Analysis was performed as follows: A custom double stranded DNA capture bait pool was used to selectively enrich the coding regions, 10 bp of flanking intronic sequences, and known relevant variants beyond the coding regions, based on HGMD® and CentoMD® for the 166 panel genes. Libraries are generated with Illumina compatible adaptors and sequenced on an Illumina platform to obtain ≥ 20x coverage depth for >99.5% of the targeted bases. An in house...
bioinformatics pipeline, including read alignment to GRCh37/hg19 genome assembly, variant calling and annotation, and comprehensive variant filtering is applied. All potential disease-causing variants, including the ones reported in HGMD®, in ClinVar and in CentoMD® are considered. Centogene has established stringent quality criteria and validation processes for variants detected by NGS. Low quality single nucleotide variants and all relevant deletion/insertion variants are confirmed by Sanger sequencing [8,9]. Consequently, we warrant a specificity of >99.9% for all reported variants. In case relevant variants are detected for a gene with available biomarker and/or enzyme activity testing at CENTOGENE, the test will be performed and included in the medical report.

RESULTS
Blood genetic analysis of the patient M.D. identified mutation existence in four genes. M.D. The following synonyms are for ATP7A: ATPase copper transporting alpha also known as Menkes' protein - MNK, CTSA (Cathepsin A, beta-galactosidase 2, beta-galactosidase protective protein, GSL, PPCA, PPGB, PPGB Human, FGF23 (Recombinant Human Fibroblast growth factor 23 protein) and PKLR (Pyruvate kinase L/R).
The ATP7A variant c.3632G>A p.(Arg1211Gln) causes an amino acid change from Arg to Gln at position 1211. It is classified as variant of uncertain significance (class 3) according to the recommendations of Centogene and ACMG (please, see additional information below). Pathogenic variants in the ATP7A gene are associated with X-linked recessive Menkes disease (OMIM®: 309400), X-linked recessive occipital horn syndrome (OMIM®: 304150), and X-linked recessive distal spinal muscular atrophy type 3 (OMIM®: 300489).
The second gene - The FGF23 variant c.61G>A p.(Val21Ile) causes an amino acid change from Val to Ile at position 21. It is classified as variant of uncertain significance (class 3) according to the recommendations of Centogene and ACMG (please, see additional information below). Pathogenic variants in the FGF23 gene are associated with autosomal dominant hypophosphatemic rickets (OMIM®: 193100).
The third gene - The CTSA gene provides instructions for making a protein called cathepsin A. Cathepsin A can act as a protease, cutting apart other proteins in order to break them down. Cathepsin A can also act as a protective protein, interacting with other enzymes to prevent them from breaking down prematurely. Based on this protective function, this enzyme is also called protective protein/cathepsin A or PPCA.
The fourth gene - The PKLR gene is active (expressed) in the liver and in red blood cells, where it provides instructions for producing an enzyme called pyruvate kinase. This enzyme is involved in a critical energy-producing process known as glycolysis. During glycolysis, the simple sugar glucose is broken down to produce energy. Specifically, pyruvate kinase is involved in the last step of the glycolytic pathway. In this step, a cluster of oxygen and phosphorus atoms (a phosphate ion) is transferred to another molecule, creating a change from phosphoenolpyruvate to pyruvate, which is then converted to ATP. ATP is the cell's main energy source. Thus, apparently, combination of gene mutations as the risk of thiazolidinedione, (2017) Clinical exome sequencing: results from 2819

REFERENCES
1. Bailey SD. Variation at the NFATC2 locus increases the risk of thiazolidinedione-induced edema in the Diabetes REduction Assessment with ramipril and rosiglitazone Medication (DREAM) study. (PMID: 20628086) DREAM investigators Diabetes care 2013. 6,12-16.

DIFFERENT GENETIC FORMS OF HEREDITARY HEMOCHROMATOSIS IN AZERBAIJAN POPULATION

Tahira Askarova

Azerbaijan Medical University.

Hemochromatoses are genetically determined or acquired forms of pathology due to excess iron in the organism [1,2,3]. There are primary and secondary forms of hemochromatosis. I type primary hereditary hemochromatosis, due to genetically deterministic hyper absorption of food iron with accumulation in liver, pancreas, myocardium, skin, endocrine glands and other tissues [4,5,6]. Secondary forms of hemochromatosis are formed in hemolytic anemia, hemotransfusion, hereditary ferrotherapy, and chronic liver diseases, especially at the stage of its cirrhosis.

According to the data of WHO, hereditary hemochromatosis (HH), the most frequent hereditary disease associated with primary iron overload, identified the I type hemochromatosis (HFE) gene, and later mutations were established in genes responsible for the development of type II-IV hemochromatosis. In Europe, the population frequency of major mutations of HH reaches 24%, in Russia 16%, and among patients with initial signs of iron overload increases to 26%. Iron accumulation has several stages: from asymptomatic period of iron overload, which is characteristic for childhood; minimal manifestations (2-3 decade of life), formed hemochromatosis (from the 4th decade of life) to multi-organ insufficiency, the main causes of mortality of untreated type I HH patients are cardiac (10-33%) and hepatic insufficiency (25-32%) portal hypertension (15%) and hepato carcinoma 23-30%. There is information in the literature that hemochromatosis is also observed in populations where blood disease is common. In Azerbaijan, as the spread of β-thalassemia is 15%, hereditary hemochromatosis can be observed in this population also.

The aim of the research. The aim of the research is the establishment of different genetic forms of inherited hemochromatosis. 29 patients with HH, homozygous heterozygous in the HH gene (17 women and 12 men) were identified aged 23 to 63 years old. the control group consists of 20 healthy people (8 women and 12 men, average age - 45 years old). A triad of clinical symptoms was observed in the patients. Iron values were examined by spectrophotometric and immunoenzyme method. The level of serum iron at patients was increased and fluctuated from 38.4 to 81.36 µmol/l (the norm is 1.87±0.17 µmol/l). The increasing constant of transferrin has consistently exceeded 60%, often reaches 157%. Level of spare iron (SF – serum ferritin, 548.4±76.94) in some cases fluctuated from 92 to 1100.5 mg/l. The level of serum transferrin equaled 271±4.2 of mg / 100. The serum iron in homozygotes was increased 1.6 times higher than in heterozygotes FSC (ferritin saturation constant) was high 1.7 times. The level of SF - in homozygotes 2.1 times was higher than in heterozygotes.

The family-geneological analysis confirmed the bearing of HH in the identified persons. An autosomal-recessive type of inheritance has been established. The phenotypic frequency of HH was 12.3%.

Hereditary hemochromatosis in the Republic of Azerbaijan

Numerous scientific studies carried out in different countries in recent years have revealed that the incidence of hereditary hemochromatosis (HH) is the most prevalent in human populations, currently not being treated, and causing serious disorders and mortality in humans.

There are many countries on the planet where hereditary hemochromatosis is spreading rapidly. Hemochromatosis can often be attributed to people living in Europe, America, Australia and sometimes in Africa and Asia. The prevalence of hereditary hemochromatosis in the European population is 10% in the heterozygous state. Genogeographical study of hereditary hemochromatosis has shown that it is widespread among the population of the Caucasus. According to the authors, the frequency of mutations C282 Y and H63D is between 1% and 24% among Caucasians. The study of hereditary hemochromatosis in the Russian Federation and Uzbekistan has not begun in the past. Earlier hereditary hemochromatosis has not been studied in our republic.

Hereditary hemochromatosis is caused by severe diseases, genetically programmed by iron deficiency in the intestines and iron loading by the body. Iron is accumulated mainly in the liver, pancreas, and heart, then damages the cells of these organs and causes the growth of the connective tissue in them. This leads to liver cirrhosis, fibrosis, and later to malignant tumors in the liver. Also, the accumulation of iron in the organs causes diabetes, other endocrine diseases, melanoderma, myocardiosplasty

Although many aspects of the disease have been studied by scientists among adults, neonatal hemochromatosis is poorly described in children, including genetic aspects of the disease, family and hereditary hemochromatosis. Due to glucose-6-phosphate dehydrogenase enzyme deficiency (11%) and high prevalence of β-thalassemia (15%) in the Republic of Azerbaijan, there are favorable conditions for the presence of carriers of two pathological genes in the presence of hereditary hemochromatosis. Also, there is little literature on the disruption of iron metabolism in the hereditary hemochromatosis in combination with structural-anomalous HbS. It is very interesting by scientifically and practically to...
study of hereditary hemochromatosis among people with diabetes, liver cirrhosis, and cardiovascular diseases. The study of hereditary hemochromatosis among the Azerbaijani population can determine the frequency of identified and theoretically expected gene frequencies.

REFERENCES

4. Аскерова Т.А., Гасанзаде Н.Ч. Возрастные особенности обмена железа у больных с наследственным гемохроматозом. Вестник проблем биологии и медицины, 2016, вып.2, том 3, с. 90-93.
THE ROLE OF ANEUPLOIDY IN THE UPREGULATION OF E2F4 AND E2F6 GENES IN BREAST CANCER CELL LINES

Nargiz Garayeva
PhD candidate, Azerbaijan Medical University. Department of Medical biology and genetics.

Strange expression pattern of stromal antigen 3 (STAG3) in breast cancer has prompted researchers to study it further as a possible breast cancer biomarker. In somatic cells there is no need for STAG3 as they divide through mitosis which has only one segregation round (segregation of sister chromatids). The expression of STAG3 gene in these cells is controlled by its negative regulators. One of them is E2F6 which is the member of E2F transcription factor family. It is encoded by the E2F6 gene located on chromosome 2p25.1 [1]. In the promoter region of STAG3 gene there is E2F site which has very high affinity to E2F6. It was proposed that to repress STAG3 gene expression in non-meiotic cells, E2F6 binds to its promoter region and recruits proteins with histone methyltransferase or histone deacetylase activity [2].

E2F4 is another member of E2F transcription family. The gene encoding E2F4 transcription factor is located on chromosome 16q22.1[1]. E2F4 also functions as a transcriptional repressor by binding to the promoter regions of its target genes in G0 and G1 phases. After that the recruitment of tumor suppressor of the pocket protein family, p130, leads to the formation of E2F4-p130 complex. This complex reduces the acetylation of histones and inhibits transcription by interacting with histone deacetylases [3].

Purpose
The purpose of current research project was to identify whether the upregulation of transcription factors E2F4 and E2F6 in breast cancer cell lines are due to aneuploidy.

Materials and methods
In this study, a control (MCF-10A) and four different human breast cancer cell lines - MCF-7, T-47D, MDA-231 and MDA-468 were used. Cells were fixed in methanol/acetic acid (50:50) on microscopic slides. Each slide was added FISH probes (Vysis CEP16[D16Z3] or Vysis CEP2[D2Z1]). Then slides were washed in 0.4xSSCT, 2xSSCT and were dehydrated sequentially in 70%, 95% and 100% ethanol series. Cell nuclei were stained by applying DAPI. Finally, cell analysis and image acquisition were performed by using OlympusBX61 fluorescence microscope equipped with Hamamatsu ORCA-ER1394 camera. FISH probe signals were scored in approximately 100 non-overlapped nuclei with clear boundaries in the chosen areas.

Results
All nuclei analyzed possessed two copies of both chromosomes in control cells. The great number of MCF-7 cell nuclei had three (46%) and four (39%) copies of chromosome 2 whereas for chromosome 16 the highest proportion of nuclei showed three copies. The number of fluorescent signals for chromosome 2 were two in 98% of tested T-47D cell nuclei. However, for chromosome 16, results were 68% with three signals and 27% with two signals. MDA-231 cell line showed four signals for both chromosomes each accounting for 93% of all nuclei examined. Copy numbers in MDA-468 cell line were very variable ranging from two to five copies, possibly showing tumor cell heterogeneity.

Conclusion
This research project showed that aneuploidy can be the key mechanism leading to the upregulation of E2F4 and E2F6, in the cell lines having more than two copies of corresponding chromosomes. However, further experiments addressed learning the other mechanisms including gene amplification and epigenetic regulation are required.

REFERENCES
İMMUNE DİAGNOSİS OF CANDLE-LİKE SYNDROME, AN AUTO-İNFLAMMATORY DİSEASE

Lala Allahverdiyeva, Naile Guliyeva

Azerbaijan Medical University, Department of Allergology and Immunology, Azerbaijan.

Auto-inflammatory diseases are genetically caused heterogeneous diseases. These pathologies are mainly associated with disorders of the mechanism of non-specific immunity. In some cases, this may occur with a violation of the regulation of specific immunity. One of these pathologies is the CANDLE-like syndrome. The disease occurs mainly in the first months and progresses. This is mainly due to the autosomal recessive mutation in the PSMB8 gene. This pathology is a chronic atypical dermatosis characterized by weight loss due to lipodystrophy, fever, joint pain, subcutaneous nodules, subcutaneous fat and muscle dystrophy.

During the first few months, the patient had a high fever, small nodules under the skin for 2 months, and then large painful nodules, erythema throughout the body, lipoatrophy, joint pain, bloating, hepatosplenomegaly.

A general analysis of blood, urine and feces, a biochemical study, blood coagulation, some hormonal tests, an extensive immunological study, and determination of auto-specific antibodies were performed during laboratory studies. Due to the results, it can be noted that the number of leukocytes, ECS, ALT, AST, QF, QQT, bilirubin and its fractions, total protein, albumin, Na, K, CL, Ca, Mg, P, uric acid, urea, sugar, creatinine kinase, ASO, reticulocyte count, prothrombin index and fibrinogen, free forms of the hormones TSH, T3- and T4 were within normal limits. ANA, Anti-dsDNA and Anti-ssDNA were negative for autoantibodies, EBV IgG and IgM were negative for specific antibodies, and Streptococc A was negative for yawning. Red blood cells, platelets, cholesterol, LDL, triglycerides, transferrin, erythropoietin, CRZ were above normal. Hb, both the relative and the absolute number of lymphocytes, Fe and its absorption, ferritin is 2.5-3 times higher than normal, HDL, creatinine and vit. D was below normal. Extensive immunological studies have revealed a decrease in IgA, IgE, CD3, CD4 and CD19, an increase in absolute IgG, CD8, CD16 / CD56, CD4 / CD8 and HLA-DR. Although genetic testing did not reveal the PSMB4, PSMB8, PSMB9, and PSMA3 genes, the patient was diagnosed CANDLE-like syndrome because all clinical signs and course of the disease resembled suppository syndrome.

Keywords: Auto-inflammatory diseases, genetic defects, CANDLE-like syndrome

REFERENCES

3. Özgür Kasapçopur., Kenan Barut., Sezgin Şahin., Amra Adrovic/ Çocukluk çağında otoenflamatuar hastalıklar/2016., p 2-10
EDITORIAL BOARD

Honorary Editors

Davit Tophuria
Tbilisi State Medical University. Head of International Students Academic Department, Associate Professor. PhD in HNA.

Nino Didbaridze
Microbiology and Immunology Department. Immunology Direction. Tbilisi State Medical University. PhD MD.

Nino Pirtskhelani
Associated Professor of Department of Molecular and Medical Genetics of Tbilisi State Medical University.

Rusudan Sujashvili
New Vision University. School of Medicine. Professor,

Tamar Giorogadze
Tbilisi State Medical University. Department of Histology, Cytology and Embryology. Assistant Professor.

Tamar Didbaridze
Tbilisi State Medical University, First University Clinic. PhD in MD.

International Advisory and Reviewer Team

Azerbaijan

Amir V. Aliyev
Ministry of Health of Azerbaijan Republic Lung Diseases Department. Guba District Central Hospital Head of Department. PhD of Medicine

Aytenok Hasanova
Azerbaijan Medical University. I Preventive Medicine Faculty. Deputy of Dean. PhD in Medical Biology.

Araz Manucher-Lalen
Associated Professor, PhD Department of Psychiatry, Azerbaijan Medical University.

Azer K. Mustafayev

Djamil Alakbarov
A researcher at the Research Institute for Lung Diseases. PhD in medicine. Azerbaijan

Jamala Mursalova
Azerbaijan National Academy of Sciences. Genetic Resources Institute. PhD BS.

Leyla I. Djafarova
Clinic “Medium” Baku. Doctor of Medical Sciences. Professor

Naila Quliyeva
Azerbaijan Medical University. Assistant in “Immunology” Program at Paediatrics Diseases Department. Docent and Academic Manager in “Allergology and Immunology” Department.

Rashad G. Abishov
Dental Implant Aesthetic Center Harbor Hospital, Azerbaijan State Doctors Improvement Institute. PhD. Azerbaijan.

Sayyara Ibadullayeva

Tariel Omarov
Azerbaijan Medical University. Department of surgical diseases. PhD in Medicine

Tubukhanum Gasimzadeh
Azerbaijan National Academy of Sciences. Institute of Dendrology of Azerbaijan NAS. Leading researcher PHD in Biological Sciences, Associate Professor.

Georgia

Eter Bukhnkashvili
Dental clinic “NGM-Innovation Dental”. The doctor-stomatologist. PhD in Medicine.

Gulnara Kiliptari
Tbilisi StateMedical University. Head of ICU department. Associate professor.

Iamze Taboridze
Scientific Center of the Humanitarian Educational University, Head, PhD in Medicine. Associate professor.

Maia Matoshvili
Tbilisi State Medical University. The First University Clinic. Dermato-Venerologist. Assistant Professor. PhD in DAPS.

Mariam Darbaidze
Davit Aghmashenebeli National Defense Academy of Georgia. The Head of Education Division. PhD in Biology.

Mariam Kharashvili
Ilia State University. Asistent Professor. PhD MD.

Nino Gogokhia
Tbilisi State Medical University. Head of Laboratory the First University Clinic. Professor.

THE FIRST INTERNATIONAL SCIENTIFIC PRACTICAL VIRTUAL CONFERENCE HUMAN GENETICS AND GENETIC DISEASES: PROBLEMS AND DEVELOPMENT PERSPECTIVES
Nino Museridze
GGRC Georgian-German Center for Reproductive Medicine, Owner and Clinical Director. The Doctor of Medicine, Full Professor.

India

Prasanta Kumar Mitra
Sikkim Manipal Institute of Medical Sciences. Department of Medical Biotechnology. PhD in Biochemistry.

Kazakhstan

Alessandra Clementi
Nazarbayev University School of Medicine. MD, GP. Assistant Professor of Medical Practice and Family Medicine
Anar Mirazagalieva
Astana International University. Vice-President. PhD in Biology.
Gulmira Zhurabekova
Marat Ospanov West-Kazakhstan State Medical Academy. Department of Human Anatomy. Associate Professor
Marina Bobireva
West Kazakhstan State Medical University named Marat Ospanov. PhD
Nuruya Kharissova
State University of Karaganda. Associate Professor of Biological Science
Zhanargul Smailova
Head of the Department of Biochemistry and Chemical Disciplines named after MD, professor S.O. Tapbergenova NAC Medical University of city Semey.

Libya

Salaheddin Sharif
University of Benghazi, International Conference on Sports Medicine and Fitness, Libyan Football Federation- Benghazi PhD in Medicine (MD)

Poland

Robert Pawel Suslo
Wroclaw Medical University, Public Health Department, Health Sciences Faculty. Adjunct Professor of Gerontology Unit. PhD MD.

Romania

Minodora Dobreanu
University of Medicine, Pharmacy, Sciences and Technology of Târgu Mureș. Faculty of Medicine. Professor. PhD in Medicine.

Russia

Alexander A. Sazanov
Leningrad State University named A.S. Pushkin. Doctor of Biological Sciences. Professor
Grigory G. Levkin
Siberian State Automobile and Highway Academy. Omsk State Transport University. PhD of Veterinary Sciences
Nikolay N. Sentyabrev
Olga Pavlova
Medic University named Rehabilitation, Doctors and Health, Professor of the Department of Morphology and Pathology, Doctor of biological sciences, physiology
Sergei A. Ostrooumov
Moscow State University. Doctor of Biological Science. Professor

Serbia

Jane Paunkovic
Faculty for Management, Megatrend University. Full Professor. PhD, Medicine

Turkey

Mehmet Inan
Turkish Physical Education Teachers Association. Vice president. PhD in Health Sciences, Physical Education and Sport Sciences
Muzaffer Sanci

THE FIRST INTERNATIONAL SCIENTIFIC PRACTICAL VIRTUAL CONFERENCE HUMAN GENETICS AND GENETIC DISEASES: PROBLEMS AND DEVELOPMENT PERSPECTIVES
Vugar Djaforov
Medical school at the University of Ondokuzmayis Turkey. PhD, Turkey.

Ukraine

Alla Oleksyuk-Nexhames
Lviv University of Medicine. Neurologyst at pedagog, pryvaty refleksoterapy. MD PD.

Dmytro Horilyk
Head of the Council, at Pharmaceutical Education & Research Center. PhD in Medicine.

Hanna Huliaieva
Institute of Microbiology and Virology, NASU, department of phytopatogenic bacteria. The senior research fellow, PhD in Biology.

Roman Lysyuk
Assistant Professor at Pharmacognosy and Botany Department at Danylo Halytsky Lviv National Medical University.

USA

Nicolai Panikov
Lecturer at Tufts University. Harvard School of Public Health. PhD/DSci, Microbiology

Rose Berkun
State University of New York at Buffalo. Assistant Professor of Anesthesiology, PhD. MD

Wael Al-Husami
Lahey Hospital & Medical Center, Nardone Medical Associate, Alkhaldi Hospital, Medical Doctor, International Health, MD, FACC, FACP

Uzbekistan

Guzel Kutlieva
Institute of Microbiology. Senior Researcher. PhD in BS.

Khurshida Narbaeva
Institute of Microbiology, Academy of Sciences Republic of Uzbekistan, Doctor of biological sciences.

Shaklo Miralimova
Academy of Science. Institute of Microbiology. Doctor of Biology Sciences. PhD in BS.
Representation of Azerbaijan International Diaspora Center in Georgia is publishing scientific papers of scientists on Website and in Referred Journals with subjects which are mentioned below:

SOUTHERN CAUCASUS SCIENTIFIC JOURNALS

Black Sea Scientific Journal of Academic Research has ISSN, E-ISSN and UDC numbering:
ISSN: 2346-8068 (Print), E-ISSN: 2346-8181 (Online), DOI: 10.36962/ALISJMSC

AGRICULTURAL, ENVIRONMENTAL & NATURAL SCIENCES

Agriculture, Agronomy & Forestry Sciences
History of Agricultural Sciences
Plant Breeding and Seed Production
Environmental Engineering Science
Earth Sciences & Organic Farming
Environmental Technology
Botany, Zoology & Biology

SOCIAL, PEDAGOGY SCIENCES & HUMANITIES

Historical Sciences and Humanities
Psychology and Sociology Sciences
Philosophy and Philology Sciences
History of Science and Technology
Social Science
Pedagogy Science
Polilology
Geography
Linguistics

MEDICINE, VETERINARY MEDICINE, PHARMACY AND BIOLOGY SCIENCES

Clinical Medicine
Prophylactic Medicine
Theoretical Medicine
Stomatology & Dentistry
Veterinary Medicine and Zoo
Drug Technology and Organization of Pharmaceutical Business
Pharmaceutical Chemistry and Pharmacology
Standardization and Organization of Medicines Production
History of Pharmacy
Innovations in Medicine
Biophysics and Biochemistry
Radiology and Microbiology
Molecular Biology and Genetics
Botany and Virology
Microbiology and Hydrobiology
Physiology of Plants, Animals and Humans
Ecology, Immunology and Biotechnology
Virology and Immunology
History of Biology
Entomology

TECHNICAL AND APPLIED SCIENCES

Applied Geometry, Engineering Drawing, Ergonomics and Safety of Life
Machines and Mechanical Engineering
History of Science and Technics
Electrical engineering, Radio Engineering, Telecommunications, and Electronics
Civil Engineering and Architecture
Information, Computing and Automation
Mining and Geodesy Sciences
Metallurgy and Energy
Chemical Technology, Chemistry Sciences
Technology of Food Products
Technology of Materials and Products Textile and Light-load industry
Machinery in Agricultural Production
History of Art
Project and Program Management
Innovative Technologies
Repair and Reconstruction
Materials Science and Engineering
Engineering Physics
Mathematics & Applied Mathematics

REGIONAL DEVELOPMENT AND INFRASTRUCTURE

History of tourism
Theoretical and methodological foundations of tourism and recreation
Tourist market, its current state and development forecasts
Training and methodological support

ECONOMIC, MANAGEMENT & MARKETING SCIENCES

Economics and Management of Enterprises
Economy and Management of a National Economy
Mathematical Methods, Models and Information Technologies in Economics
Accounting, Analysis and Auditing
Money, Finance and Credit
Demography, Labor Economics
Management and Marketing
Economic Science

LEGAL AND POLITICAL SCIENCE

Theory and History of State and Law
International Law
Branches of Law
Judicial System and Philosophy of Law
Theory and History of Political Science
Political Institutions and Processes
Political Culture and Ideology
Political Problems of International Systems and Global Development

CONFERENCE NEWSLETTER

MULTIDISCIPLINARY JOURNAL
Clinical Medicine
Prophylactic Medicine
Theoretical Medicine
Stomatology & Dentistry
Veterinary Medicine and Zoo
Drug Technology and Organization of Pharmaceutical Business
Pharmaceutical Chemistry and Pharmacology
Standardization and Organization of Medicines Production
History of Pharmacy
Innovations in Medicine
Biophysics and Biochemistry
Radiology and Microbiology
Molecular Biology and Genetics
Botany and Virology
Microbiology and Hydrobiology
Physiology of Plants, Animals and Humans
Ecology, Immunology and Biotechnology
Virology and Immunology
History of Biology
Entomology

http://sc-media.org/ambiance-in-life-isjmsc/