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14th Balkan Congress of Human Genetics
& 9th Rare Disease SEE Meeting 2023

Skopje, October 05-07, 2023

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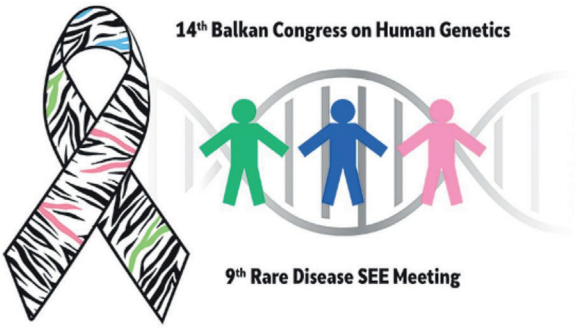

ABSTRACT BOOK

14th Balkan Congress of Human Genetics and 9th Rare Disease SEE Meeting

"Genetic Diseases from Diagnostics to Prevention and Therapy"

October 05-07, 2023
Hotel "DoubleTree by Hilton"
Skopje, North Macedonia

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Disclaimer

This abstract book has been produced using author-supplied copy. Editing has been restricted to some corrections of spelling and style where appropriate. The organizing Committee assumes no responsibility for any claims, instructions, methods or drug dosages contained in the abstracts.

TABLE OF CONTENTS

Organizers:

Македонско здружение за хумана генетика
МЗХГ
Macedonian Society of Human Genetics (MSHG)

Macedonian Society for Rare Diseases

LIFE with CHALLENGES
Life is a puzzle, collect it.

РЕТКО Е ДА СИ РЕДОК
RRALLË ËSHTË TË JESH I RRALLË
RARE IS TO BE RARE

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Welcome Address

Dear Colleagues, Friends, Guests,

It is indeed a great honor and pleasure for me to extend you all a very warm welcome, on behalf of the Organizing Committee of the 14th Balkan Congress of Human Genetics and the 9th Rare Disease SEE Meeting. This joint conference brings together leading scientists, researchers, clinicians, and industry professionals from the Balkan region and wider to share their latest findings and developments in the fields of genetics and rare diseases and to exchange experiences.

The conference bears the general designation "Genetic diseases – from diagnostics to prevention and therapy" and covers various areas related to the stated topics in rare diseases as well as in complex diseases influenced by genetic factors. The conference aims to promote knowledge exchange and collaboration between experts in the field, and to provide a platform for discussing the latest advances and challenges in genetic research. It is primarily dedicated to professionals in the field of human genetics, including but not limited to the fields of pediatrics, neurology, cardiology, hematology, oncology, obstetrics and gynecology, and rare diseases.

The program is arranged in a multidisciplinary manner in order to allow delegates to both explore issues pertinent to their own area of interest and to interact with other professionals outside of their specialty. The conference features plenary lectures given by renowned European experts, invited lectures given by leading scientists from the Balkan countries, oral and poster presentations, as well as satellite symposia providing ample opportunities for attendees to network, learn, and share their research. In addition, there is a rich commercial exhibition, where the latest genetic technologies, products, and services are showcased.

The abstracts of the conference have been published in a Supplement of the Balkan Journal of Medical Genetics, an international journal published by the Macedonian Academy of Sciences and Arts, which is cited in all major medical and scientific databases, including PubMed, Web of Science and others.

The conference is dedicated to the patients and especially to those with rare diseases. Indeed, patients' organizations for rare diseases from our country have been our partners in the organization of this event.

To conclude, I would like to thank each and every one of you for attending this conference. I am confident that you will have an exciting, stimulating, and productive meeting. Nevertheless, I also hope that you have planned some extra time to enjoy our culture, food, tradition and hospitality.

Добредојдовте! Welcome!

Prof. Dr. *Dijana Plaseska-Karanfilska*
President of the Organizing Committee

General Information

Venue:
Platinum Conference Room
Hotel "DoubleTree" by Hilton
Blvd "ASNOM" 17,
1000 Skopje
North Macedonia

Registration desk:

Reception desk is organized in front of the Platinum Conference Room (1 and 2) at the Hotel "DoubleTree" by Hilton and will be open for information and registration from 10:00am 05.10.2023

Language:

The official language of the Congress is English.

Oral presentations

Facilities will be available for Microsoft PowerPoint (ppt or pptx) slides, in 16:9 format. Speakers are kindly requested to hand their presentations to the designated technician in the Conference room in the morning or during the breaks, but at least 1 hour before the start of the session.

Please, take in consideration the allocated time for effective presentation of your presentation.

Poster presentations

Posters will be presented on three 65" TV Monitors, according the detailed Program in two Poster sessions. Authors must be present at the boards for the duration of their scheduled poster session. Posters will be on display for the duration of the Conference (Thursday, October 5th, 2023 at 12:00, till Saturday, October 7th, 2023 till 15:00).

Registration Fee

Registration fee covers the Program, access to the Scientific sessions, Opening Ceremony, Get together Party, Coffee brakes, Lunch.

PDF version of the Abstract bookis available on the Event web-site.

Certificate for Attendance will be available at the event web-site (https://mzhg.org.mk) with appropriate log-in.

PP-06

POTENTIAL NEW GENES INVOLVED IN CYSTIC FIBROSIS PHENOTYPE

Jelena Kusic-Tisma, Dusan Usjak, Martina Mia Mitic, Aleksandra Divac Rankov

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Background: Cystic fibrosis (CF) is autosomal recessive disorder characterized by chronic respiratory problems and poor growth. CF is caused by defect in transmembrane conductance regulator (CFTR) protein. CF is diagnosed by sweat chloride analysis (>60 mmol/L) with the identification of two CF-causing variants of *CFTR* gene. With a longstanding history of *CFTR* gene analysis, our laboratory identified several patients with elevated sweat chloride and clinical manifestations of CF in whom no CF-causing mutations were detected after sequencing of whole coding region and testing for large insertion/deletion of *CFTR* gene. In order to elucidate genetic background of conditions that mimic CF we performed whole exome sequencing (WES) in two such patients.

Methods: Library preparation was done using DNA nanoball technology. Produced fastq files were mapped to hg38. VCF files were generated using GATK and annotated with InterVar and AnnoVar tools. Variants filtering for disease relevance was done using the following criteria: QC, GnomAD Allele Frequency, Functional consequences and phenotype-genotype relationship.

Results: *CACNA1H* and *MUC5B* genes were found to be impaired in both patients. Similar number of variants predicted to impair protein function were detected (27 and 25) in each patient. Loss of function variants were found in 7 and 11 genes, respectively.

Conclusion: Further assessment of selected variants will clarify their functional effect and relevance for the patient's clinical phenotype. WES analysis will help identify genetic aspects of disease and assist in optimal patient management in about 0.01% of patients with elevated sweat chloride and high clinical suspicion of CF that do not carry any CF-causing variants.

Keywords: *cystic fibrosis, WES, variant assessment, patient management*

Topic*: Rare diseases*