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& 9th Rare Disease SEE Meeting 2023

Skopje, October 05-07, 2023

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EDITORIAL OFFICE
Macedonian Academy of Sciences and Arts, Bul. Krste Misirkov 2, POB 428
1000 Skopje, Republic of North Macedonia.
Phone: +389 2 3235 411, fax: +389 2 3115 434, E-mail: bjmg@manu.edu.mk
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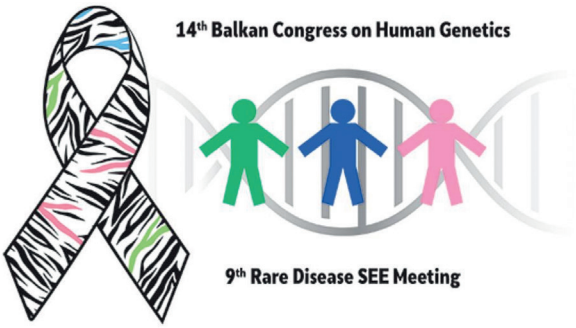

ABSTRACT BOOK

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.

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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

ВИЛСОН МАКЕДОНИЈА

CONCEPT marketing communications

Sponsors:

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GENESIS pharma

BI EL SINCE 1992

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Matevska Nadica, North Macedonia

Scientific Committee:

Plasheska-Karanfilska Dijana, North Macedonia
Tasic Velibor, North Macedonia
Sukarova Angelovska Elena, North Macedonia
Dimovski Aleksandar, North Macedonia
Petlichkovski Aleksandar, North Macedonia
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Streata Ioana, Romania
Syrrou Maria, Greece
Trajkova Slavica, Italy
Xhetani Merita, Albania

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.

OP-02

OUTCOME OF A SERBIAN PILOT INITIATIVE: SPINAL MUSCULAR ATROPHY NEWBORN SCREENING OVER A 16-MONTH PERIOD

Milos Brkusanin[1], Nemanja Garai[1], Jelena Karanovic[1], Matija Trickovic[1], Dimitrije Nikolic[2], Tamara Sljivancanin Jakovljevic[3], Aleksandra Dimitrijevic[4], Kristina Jovanovic[5], Dusanka Savic-Pavicevic[1]

[1] Centre for Human Molecular Genetics, University of Belgrade-Faculty of Biology, Belgrade, Serbia

[2] University of Belgrade-Faculty of Medicine, Belgrade, Serbia

[3] Department of Neonatology, Obstetrics and Gynaecology Clinic Narodni Front, Belgrade, Serbia

[4] Department of Gynecology and Obstetrics, Faculty of Medical Sciences, University of Kragujevac, Kragujevac, Serbia

[5] Neurology Department, University Children's Hospital, Belgrade, Serbia

Presenting author e-mail: milosb@bio.bg.ac.rs

Background: Spinal muscular atrophy (SMA) is the prevalent genetic cause of childhood mortality. Pioneering treatments yield utmost advantages only within the presymptomatic phase, underlining the significance of newborn screening.

Materials and methods: In 2022, the Centre for Human Molecular Genetics initiated a pilot study of the newborn screening for SMA, working closely alongside the University Children's Hospital Tirsova and Association SMA Serbia. The aim was to lay the foundation for the implementation of statewide newborn screening for SMA in Serbia by conducting screening for ~8000 infants from the Obstetrics and Gynaecology Clinic Narodni Front over the course of a year.

Results: In the initial year, 6950 newborns underwent testing, revealing SMA in two unrelated infants and in an asymptomatic 16-month old sibling of the first newborn. All three children received therapeutic interventions in <1 month from birth. To date, they have exhibited no signs of SMA, and there have been no false-negative outcomes among the newborns who tested negative during the screening.

As frontrunners in this field in Serbia, we orchestrated harmonized efforts across various tiers of healthcare, established screening and diagnostic algorithms and follow-up protocols. In the second year, we included a maternity hospital beyond Belgrade, introducing sample shipping via mail and extending screening accessibility to a greater number of infants. This resulted in 9800 infants undergoing testing within 16 months. Currently, we are actively preparing for the official incorporation of newborn screening for SMA into the national screening program.

Conclusions: Timely detection and treatment can transform SMA into a manageable condition.

Keywords: *Newborn screening, Spinal muscular atrophy, Pre-symptomatic diagnosis, SMA prevention, SMN1*

Topic: *Rare diseases*