Macedonian Academy of Arts and Sciences (MASA), Skopje

6th RARE DISEASE IN SEE MEETING, SKOPJE, MACEDONIA
(EAP, UEMS Section of Paediatrics)

Saturday, November 11, 2017

Organizing Committee:

Momir Polenakovic, President
Felix Unger, Co-President
Zoran Gucev, Secretary
Nada Pop-Jordanova
Ilija Filipche
Zivko Popov
VeliborTasic
Katarina Stavric
Vesna Aleksovska

FRIDAY, NOVEMBER 10, 2017

20.00 Get together, Hotel Kontinental Skopje

AGENDA

SATURDAY, NOVEMBER 11

Rare Diseases in SEE

Moderators: Nada Pop-Jordanova, Zoran Gucev

09.00-09.15 **Welcome and opening**

Session I

09.15-09.25 **Momir Polenakovic, Skopje, Macedonia**
Introdcutory remarks

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09.25-09.40 **Vesna Aleksovska, Anja Bosilkova Antovska, Skopje Macedonia**
National plan for rare diseases an regulative for rare diseases in Macedonia - challenges and solutions

09.40-10.05 **Ria Schönauer, Leipzig, Germany**
Intrafamilial variability due to genetic modification in ADPKD

10.05-10.30 **Mirko Spirovski, Skopje, Macedonia**
Rare-diseases genetics in the era of next generation sequencing: Single Center experience

10.30-10.55 **Johannes A. Mayr, Salzburg, Austria**
Next generation sequencing reveals new treatable diseases. Are we entering a new era of preventive medicine?

10.55-11.20 **Zoran Gucev, Skopje, Macedonia**
Rare diseases, new genes, molecular mechanisms and treatments

Discussion

11.20-11.45 **Coffee break**

11.45-12.10 **Oliver Bartsch, Mainz, Germany**
Clinical update on preimplantation genetic diagnosis

12.10-12.35 **Martin Magner, Prague, Chech Republic**
Mucopolisaccharidoses

12.35-13.00 **Velibor Tasic, Skopje, Macedonia**
Pediatric nephrology in the next generation sequencing era

13.00-13.25 **Ramush Bejiqi, Prishtine, Kosovo**
Klippel-Feil syndrome associated with congemital heart disease

Discussion

13.25-14.10 **Lunch and e-poster session**

Session II

Moderators: Mirko Spiroski, Velibor Tasic

14.10-14.35 **Ruthild Weber, Hanover, Germany**
Genetics of renal malformation: new insights through next generation sequencing.

14.35-15.00 **Dieter Haffner, Hanover, Germany**
Complications and management of X-linked hypophosphatemia

15.00-15.25 **Julia Hoefele, Munchen, Germany**
Impact of genetic modifiers in female patients with Alport syndrome

Discussion

15.25-15.50 **Coffee break**

15.50-16.10 **Milosevic Danko, Zagreb, Croatia**
Atypical hemolytic-uremic syndrome (aHUS)-Croatian experience

16.10-16.30 **Adrijan Sarajlija, Belgrade, Serbia**
Clinical spectrum of mitochondrial disorders in childhood

16.30-16.50 **Vjosa Kotori, Prishtine, Kosovo**
Hyperinsulinemic hypoglycemia

Discussion

Conclusion remarks

Zoran Gucev, Velibor Tasic, Momir Polenakovic

Venue

Macedonian Academy of Arts and Sciences
Bul. KrsteMisirkov, No.2, Skopje
Republic of Macedonia

Language of the Meeting: English

Certificates of Attendance will be provided by Prof. V. Tasic