REPORT ON THE 8th RARE DISEASE IN SEE MEETING

(EAP, UEMS Section of Paediatrics)

Hotel Sileks, Ohrid, Republic of North Macedonia

23rd to 25th September 2022

Organizing Committee:

President: Zoran Gucev, Aspazija Sofijanova, Velibor Tasic; Aleksandra Janchevska, Secretary; Todor Arsov; Elena Shukarova Angelovska; Marina Krstevska Konstantinova; Vesna Aleksovska

International Scientific Committee:

Chair:Velibor Tasic Skopje, North Macedonia; Zoran Gucev, North Macedonia

Members: Adrijan Sarajlija, Belgrade, Serbia; Danko Milosevic, Zagreb, Croatia; Jasmina Comic, Munchen, Germany; Constantinos Stefanidis, Athens Greece; Maria Gaydarova, Sofia Bulgaria

The congress opened with a press conference and statements from relevant stakeholders. Over 120 people attended the meeting, including health professionals, researchers, patients and families with rare diseases, jounralists, representatives from institutions, and representatives from pharma companies.

- **Mr. Bekim Sali, Minister of Health, North Macedonia:** The Minister talked about the progress made in the rare disease program in North Macedonia and the future steps to improve life of patients with rare diseases in our country.
- **Mrs. Aspazija Sofijanova, University Children's Hospital:** The director of the children clinic, talked about the need of cooperation to improve neonatal screening and diagnosis and the importance of all stakeholders working together towards solutions to the challnges that we have at the present.
- **Ms Vesna Aleksovska, Chair of the rare disease association Life with challenges:** As a patient advocate, Vesna, talked about the current problems and challenges that families face, focusing on the problems of procurement of medicines and access to diagnosis and screening.

There were lecturers from UK, Germany, Portugal, Croatia, Serbia, North Macedonia, Bulgaria, and Greece. We managed to cover many different rare diseases such as: Phenylketonuria, Gaucher Disease, Alport Syndrome, Achondroplasia, Alagille Syndrome,

Spinal Muscular Atrophy, Muscular Dustrophy, MPS 2 – Hunter syndrome, Hyperoxaluria, Wilson disease and more was covered in the poster presentations on Sunday. ***All lectureres biographies and abstracts are included in the abstract book that is attached to this report.***

It is important to mention that we also started a cooperation with the Association of General practitioners to work together on improvement on diagnosis, through education, workshops, informative materials and other activities, starting with the attendnce of around 20 doctors at this meeting. The doctors agreed that it would beneficial not just to have lectures from doctors, but to also hear patient and family stories. These activitites will be organized next year (we hope to organize 4 workshops in the period from April to November, 2023).

The conference started with the lectures of: Zoran Gucev, Skopje, North Macedonia (PIK3CA-related overgrowth spectrum (PROS): New treatments), **Klaus Mohnike, Magdeburg, Germany, (**Experiences in pharmacological treatment of achondroplasia dwarfism with vosoritide), **Elena Sukarova Angelovska, Skopje, North Macedonia** (Phenylketonuria (PKU) – Genetic variations and their clinical presentations in PKU), **Júlio César Rocha, Lisbon, Portugal, (**How to implement basic PKU management practices toward the best standar of care), **Aspazija Sofijanova, Skopje, North Macedonia (**Introducing inovative therapy and experince with SMA in North Macedonia, will genetic therapy be final solution?), **Dimitrije Nikolić, Belgrade, Serbia (**Neonatal screening for SMA in Serbia – First Results), **Tanja Loboda, Ljubljana, Slovenia (**Duchenne muscular dystrophy - from early diagnosis to novel therapies). **Constantinos Stefanidis, Athens, Greece (**Epidemiology of hyperoxaluria type 1 (PH1), **Maria Gaydarova, Sofia Bulgaria (**Status of PH1 in Bulgaria and patient cases).

Gaucher disease session

At the meeting we had a special session dedicated to Gaucher disease where we had the opportunity to listen to the lectures of Prof Timothy Cox (Gaucher disease matters), Dijana Plasheska Karanfilska (Genetics of Gaucher disease) and Goran Chuturilo (Importance of diagnostic of rare diseases with enzyme and genetic testing).

The lecturers focused on diagnosis as in the past 15 years we haven't had children diagnosed with Gaucher disease which is a problem that needs resolve as soon as possible.

Families with Gaucher disease from North Macedonia attended the meeting and had opportunity to meet the experts and talk about current challenges and possible solutions (photos from them are not included as we do not share patients photos and information).

Gaucher families also had a spearate patient meeting one month previously (20-21 August, Struga, North Macedonia), where they talked about what they need in the future to improve treatment and management of Gaucher disease in our country.

All of the conclusions from the family meeting including requests to improve management of the diseases and with that improve quality of life of patients and their families, were sent as a request to the University Hematology Clinic in Skopje, and unfortunately all of our requests were denied without a solution. We will continue to advocate further for improvement of the quality of life of families with Gaucher disease.

The conclusions from the family Gaucher meeting were the following:

- Need for education of family doctors (GPs) for recognizing symptoms of Gaucher disease and referring patients to the department of hematology in the University children clinic, or the University hematology clinic (depending on age of the patient).
- Need for clear guidelines on Gaucher disease management and follow up (the Children clinic already adopted one from UK).
- Taking in consideration the previous conclusion, there is also a need to establish which control tests need to be done each year for each patient, because now some patients had CT, DEXA scan and ultrasound each year, some had none for 2 years. The only constant is the blood control test that patients do every 2 weeks which is not necessary as once in 3 or 6 months would be enough to follow up the patients.
- Need for a proper treatment space for the patients at the hematology clinic (At the moment all patients are receiving treatment together in a room with mostly cancer patients which has proven to be depressing and sad for the patients and the family members. Also, the waiting time for administration documents and receiving treatment is usually from 4 to 6 hours. Additionally, there is only one bathroom for men and women and the door can't be closed, and it is usually in a very bad condition).
- Need for implementation of a home treatment program or at least decentralization of receiving treatment in other cities in North Macedonia (The travel time for some patients is 4 hours in one direction, which means that patients need to take 1 day off work to receive treatment and travel 8 hours a day. There are regional hospitals that can do treatment for patients, and also medicine distributors agree to provide cold chain to transport the medicines every 2 weeks where is needed).

The meeting closed with the lectures of: **Adrijan Sarajlija, Belgrade, Serbia** (Treatable inherited metabolic diseases), **Jasmina Comic, Munich, Germany** (Genetics of Alport syndrome-Munich experience), **Aleksandra Janchevska, Skopje, North Macedonia** (The overgrowth syndromes – the story of a patient with Beckwith Wiedemann Syndrome (BWS)), **Sonja Bojadzieva, Skopje, North Macedonia** (Wilson's Disease in Children- diagnostic aspects and therapy), **Aco Kostovski, Skopje, North Macedonia** (Allagile syndrome – new treatment possibility).

On Sunday morning we had a poster session, where a lot of medical students had the opportunity to present and get feedback from the medical professionals. There were also poster presentations by doctors with focus on case studies. We always cooperate with medical students, as they are the future and education for rare diseases is necessary to start as early as possible, so they are aware of the rare diseases that occur in North Macedonia and the possibilities of management and treatment.

At the meeting we also had an opportunity to realize an Alport meeting for the families to have an opportunity to meet with experts in the region. This meeting went parallel to the main session after the lunch break. It is never easy to organize a meeting for a disease where research is still going on and there is no current treatment. This is the second time we are managing to support such meeting by using existing resources.

Lecturers at this meeting were: **Gordana Loleska, Skopje, North Macedonia** (Alport Macedonia – Who are we?)**, Ana Momirovska, Skopje, North Macedonia** (Diagnostics of COL4A diseases in North Macedonia from the very beginning)**, Nora Abazi Emini, Skopje, North Macedonia** (Type-IV-Collagen-Related nephropathies in North Macedonia)**,Velibor Tasic, Skopje, North Macedonia** (Extrarenal features of COL4A nephropathies), **Jasmina Comic** (The Munich Microhematuria Project), **Constantinos Stefanidis** (COL4A nephropathies in Greek children)**, Nikola Gjorgjievski, Skopje, North Macedonia** (Alport syndromre-when should one consult nephrologist), **Marija Gaydarova Sofia, Bulgaria**, (COL4A Nephropathies in Bulgaria), **Danko Milosevic, Zagreb, Croatia** (COL4A nephropathies in Croatian Children), **Todor Arsov, Skopje, North Macedonia** (Challenges in genetic counselling for Alport syndrome in the age of genomic medicine).

We are very happy that this meeting provided safe space for all stakeholders and interested parties to share information, best practice, knowldege, education and also to start cooperation and work together on projects that will support improvement of early diagnosis, early access to treatment and adequate management of rare diseases, with better quality of life for the patients and the families.

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