MINUTES OF THE RARE DISEASES WORKING GROUP

SPRING MEETING 2021

LARNACA, CYPRUS / VIRTUAL JULY 2, 2021

17.45 - 18.30 CY / 16.45 - 17.30 CET

1. Approval of Agenda – The Agenda has been approved

2. Approval of the Minutes of the Previous Rare Diseases Session at the 2019 Winter Meeting – The Minutes were approved.

3. Rare Diseases in frame of Universal Health Coverage (click here)

The survey was conducted among a network of paediatricians, on issues of the directive on Universal Health Coverage of the United Nations, September 2019. We questioned "What could be the least the European paediatricians could do to realize the goals of the UHC? 80 people from the network were contacted and 48 responded. 29 Countries were covered, it has extended out of Europe. Note that 52% are tertiary care paediatricians. Most of the respondents (76%) come from high economy countries.
There were 10 questions. What would be a global action to be necessary in the frame of rare diseases? Rare diseases are only mentioned once in the health coverage directive. Number 34. Most respondents agreed diagnostics should be accessible regardless of ability to pay. There are big differences in countries. We must first recognize the child in practice before diagnosis can be achieved. Children living with the diagnosis autism, developmental delay, prolonged neonatal icterus, abnormal features, failure to thrive may very well have a **rare condition** with specific health risks and treatment. Any child could have a rare disease regardless of other disabilities. We can see these abnormalities anywhere in our practice, tertiary, or primary care. One can find diagnostic features of the child from the internet. Diagnostics features can be found even if it not in the book yet. When diagnostics become available you can conduct chromosome studies. Chromosome study: 12 months old girl with reduced growth in length and weight and minor dysmorphic features > Array CGH > Materials: EDTA blood 2x 3-6 ml > result term: 5 weeks Resulted in diagnosing a moziac duplicate chromosome 12q12-q13.2. DNA panels 2 year old girl with a developmental delay > Exome panel developmental delay (1338genes) > Materials: EDTA Blood, isolated DNA > Result term regular: 3 months/ rapid: 15 workdays:Resulted in diagnosing a mutation in NAA15
Soon, there are DNA panels which can be done. From primary care, mutations are found in these genes. What to do when a diagnosis is made? Some syndromes caused by mutations are not found in NELSON's book or in the European Masterclass. Most recent guidelines can be found on Orphanet.
Diagnostic criteria might change. DNA panels may be a solution in future. With a little blood transferred from Sri Lanka to Germany, enabled this diagnosis, a diagnosis of a rare condition was established.
Regarding COVID and the risk
Promoting primary care will decrease child mortality and increase access to health services.
Training would be the most helpful. yEAP can assist with how the training should happen. What actions are necessary? No one responded. We should support primary care practice in the lower- and middle-income countries. Disabled children are not always very well looked after in their societies.

•As most rare disease present in childhood.

•Paediatricians, at the **hospital as well as in primary care in the community**, to recognize and take are of children with rare and disabling conditions.

•The **EAP's 'Core Training' syllabus** resonates with **Universal Health Coverage**, the rights of the child and the rights of the disabled.

•The first European Board of Paediatrics Exam took place in 2020, and two further examinations are planned in 2021.

Increase **global awareness**, international solidarity, international collaboration, and action towards the achievement of the universal health coverage (UHCnr78)
No actions are necessary: 0%
Engage in high quality, affordable, sustainable, and sematic digital child health: 68%
Support families in their fundamental needs: 68%
Support primary paediatric care with an emphasis on care in rural and distant areas: 84%
Identify the specific needs of poor and vulnerable children: 79%
Establish a global paediatric network to support the sustainable development goals of the UHC: 87%

Due to the lack of affordability and availability of diagnostic facilities and therapies in middle -let alone lower -economy countries, chronically ill and disabled children are deprived of receiving accurate diagnosis or proper therapy.
Unless deliberate efforts are made to reach these children, they face preventable morbidity and death without having a chance to profit from advances in medical and digital science.
The implementation of a sustainable digital child health system, in line with the WHO recommendations on digital health as well as the UHC, requires support of policy makers.
Further research should include identification of more specific actions: "to support primary paediatric care with an emphasis on care in rural and distant areas and support the specific needs of chronically ill and disabled children".
Discussion: Expense of medication due to the limitation of access. Disruptions due to global access of supplies. Critical medications have been running out. Pharmaceutical companies should be encouraged to join the conversation.
Investment in rare diseases has been boosted as a strong economic development. Pharmaceutical companies have a monopoly. Although we want the investment from companies, we should think about strategies that treatment is expensive. The rights of the children are universal. This topic will be addressed in the December meeting.
Courses for trainees are essential to refer for a diagnosis.
Do insurance companies cover the cost of the DNA panel? It depends which country. Not pediatricians can refer to panels for first line of diagnosis.

4. Towards a global digital child health record with FHIR: get involved! (click here)

Children are being seen everywhere in the world. Growth and development from 0-4 years, even in Africa. There is some kind of standard. The AAP recommends for primary is measuring heads, vaccinations, things that are all done in primary care.
Everywhere in the world, we gather data, growth, and development. The data can be transformed in coding, and then shared between professionals.
A small set of global health child records. For children aged 0-4. If we are all connect to HL7/ FHIR, our codes can be put into the digital system. The codes are defined.
These files can be transferred to a personal health record. There is a list of coding which is presented. If you include all the other coding, we have a more holistic system.
When we take blood from a child, it is sent to a laboratory, they're more or less the same across the world. The coding is based on a variety of tests. It can go up to the genetics. All of this can be integrated into one digital system. How this works, you see the coding in internet language, usually when you're on the back end. Vaccination and other physical exams can be done. The advantage is that the same data is collected and can be shared on the database and integrated with different other tools. This is to contribute to universal health coverage.
The rare disease group was started in 2010. The Netherlands government is supporting the HL7.
Adamos expressed although this is an ambitious project, it is very possible in the near future.
Discussion: who would be responsible for putting the codes into the system? IDC10 coding for inpatients. The coding takes time and is quite complex. If all together, creating a link with the coding. Such an application interface can be shared globally. All electronic records can be connected. The database can be created and offered as an API.