Dear all,
this is a brief report of the SSIEM 2016 held in Rome, in which our vounteer Maria Monticelli had the honour to attend and represent CDG patient voice.
Please note that this is not supposed to be a detailed report including scientific data but a summary of what has been discussed in the field of CDGs.

In summary:

European Reference Network on Hereditary Metabolic Diseases (MetabERN)

First of all, on Monday 5th September there was a meeting focused on the **European Reference Network on Hereditary Metabolic Diseases** (MetabERN).
The MetabERN is supposed to be the European Reference Network for the Metabolic diseases. The MetabERN is responding to the call for the establishment of European Reference Networks (ERNs) launched by the European Commission as laid down in the Directive on the patients' rights in cross-border healthcare[1]. For more information about MetabERN visit HERE.

The European Reference Networks are growing and spreading, and it would be absolutely great having one for metabolic diseases. A public consultation on the implementation of European Reference Networks (ERN) was done under the framework of Article 12 of Directive 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare. For more information please visit:

- The policy framework for ERNs available HERE
- What Is a European Reference Network HERE
- Information about advances on ERNs that is available HERE

Importantly, EURORDIS developed **European Patient Advocacy Group** (EPAG) for each ERN disease grouping. These ePAGs bring together elected patient representatives from EURORDIS member organisations and ensures that the patient voice is heard throughout the ERN development process. The CDG community is currently represented by the Portuguese Association for CDG (APCDG) and Dr Rita Francisco CDG Patient Advocacy Group Manager (currently researcher among APCDG).

There are different steps to be approved as official ERN. The application for our Network has been done on the 21st June 2016. The first screen, for the eligibility, ended up with a positive result. The second screen will be done from a technical committee, in October. Let's hope it will be positive too!

The MetabERN will be composed by a Multidisciplinary Team (MDT), with 1681 physicians (from 18 countries) taking care of more than 40000 patients (1/3 adults and 2/3 pediatric) with 7 disease categories. CDG is one of these categories. **Patients with a CDG are almost 1% of the total.**
The aim of the MetabERN will be focused on:

- prevention and screening
- diagnosis/new diseases diagnosis
- management
- epidemiology
- education
- virtual counselling

[1] http://www.brains4brain.eu/the-european-reference-network-for-hereditary-metabolic-diseases-metabern-is-underway/

- dissemination
- clinical trials
- patient empowerment
- research

During the meeting, the importance of the patients' associations was highlighted by Lut de Bare, the President of the Belgian Patient Organization for Metabolic Diseases.
In the next months, a lot of work will be done to share the data ad summarize them in order to establish priorities. Hoping that the approval will arrive soon, the date for the 3rd Conference on European Reference Networks will be 9-10 March 2017, to be held in Vilnius (Lituania).

On Tuesday 6th September, the opening ceremony of the SSIEM Symposium was masterfully opened by Pf. Eva Morava.

CDG SESSIONS DURING SSIEM CONFERENCE

The CDG session was held on Thursday 8th September.

Pf. Jaeken, giving an update lecture titled "What is new in CDGs", elucidated novel types of CDGs discovered recently or new phenotypes associated with known mutations, such as the XILT2-CDG, the EXT2-CDG, and the PGM3-CDG (more information HERE). He also caught the opportunity to cite the Portuguese Association for CDG and to thank them for their tremendous input.

Dr. Lefeber, from the Netherlands, gave a talk about "A novel group of metabolic disorders due to tissue-specific defects in V-ATPase assembly", explaining how they went through the diagnosis of a large group of CDG-II patients and discovered 4 novel gene defects. The effects of these gene defects are a (at least partial) explanation for the tissue-restricted disease symptoms.

Dr. Yuste-Checa, from Spain, talked about "Toward a folding therapy for PMM2-CDG", explaining how, in her research activity, she focused on the combination between pharmacological chaperones and proteostasis regulator. Protein misfolding is an important disease mechanism in PMM2-CDG. Helping PMM2 to acquire and maintain the folded structure is a most promising research line for this disease. For more information related to this work read HERE.

The lecture of Dr. Peanne, from Belgium, focused on "MAGT1-deficiency: new insights into a controversial protein with a key role in N-glycosylation". MAGT1 (Magnesium transporter protein 1) is a new gene candidate for CDG because this protein is an important part of the glycosylation machinery. Two patients were discovered with mutations in this gene. The link with magnesium transport is still enigmatic and will be further studied. More information read HERE.

Finally, Dr. Park from Germany gave a talk about "SLC39A8 deficiency is a novel treatable disorder of manganese metabolism and glycosylation". He explained the way his research group identified a patient who revealed sensitive to a treatment with manganese and threw the challenge of identifying new responsive patients. For more information about SLC39A8 deficiency visit HERE.

CDG BOOTH AT SSIEM CONFERENCE

Our booth has been accessible during the whole week, with a lot of posters explaining all the work we are doing in research and in education. Many people came to congratulate for the work and to ask for information.

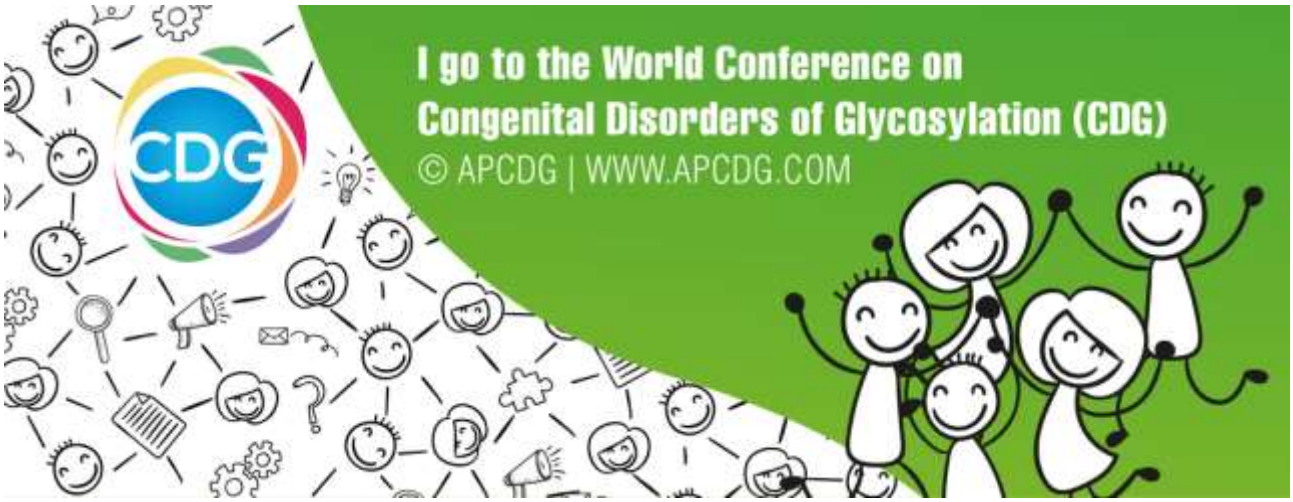
Moreover, the work titled "Liver involvement in Congenital Disorders of Glycosylation – Literature review" was accepted and exposed as a scientific poster. Maria Monticelli (me) and Pf. Jaeken had the opportunity to explain the work we have done to several interested people.

The SSIEM Annual Symposium 2016 has been a great experience and it gave to our community the opportunity to share knowledge, experiences and wishes. The research in CDG is yielding a lot of results, and even if we are still at the beginning, the cooperation between professionals and families is really promising!

I wish to thank the APCDG for trusting me in representing the CDG community at the CDG booth.

Just to conclude, I remind you that the next International Scientific CDG Symposium will take place in Leuven, Belgium, on 13-14 July 2017, and the "**3rd World Conference on Congenital Disorders of Glycosylation (CDG) for families and professionals**: United shaping the future for CDG"on **15 and 16 July 2017** also in Leuven, Belgium. For more information visit HERE.

Let's meet in Leuven (Belgium)!



Visit the website dedicated to "3rd World Conference on Congenital Disorders of Glycosylation (CDG) for families and professionals HERE.