

## Lay Language Summary of the Article “Public and patient involvement in needs assessment and social innovation: a people-centered approach to care and research for congenital disorders of glycosylation”

Propose title for the lay summary:

### **A community-centred approach to identify needs and promote innovation in congenital disorders of glycosylation (CDG) care and research**

You can access the article’s abstracts [HERE](#).

To get a free full copy write to [sindromeCDG@gmail.com](mailto:sindromeCDG@gmail.com)

### **What is the Think Tank methodology?**

Also called think factory, it is a gathering of a group of experts with the objective of analysing and discussing complex problems and suggesting possible solutions.

### **Take home messages**

- The main objectives of this work were: **1)** To assess the needs of the CDG community; **2)** To create solutions as a community to the unmet needs identified. Basically, apply the Think Tank methodology to pinpoint the needs and simultaneously develop responses for the problems identified;
- 48 participants, including 18 patients/family members, 7 health care professionals, 7 researchers and 16 people combining several of these roles from 20 countries distributed across the five continents were randomly enrolled in the study;
- Participants defined as areas of major concern: Information, health care, psychosocial support and representation in care and research;
- Solutions proposed by the participants were civil society, care-related and digital innovations;
- Innovative aspects of this study were the inclusion of both professionals and families, allowing also the establishment of needs, priorities and solutions at the international level, consequently permitting a more comprehensive vision and overview of this community in a face-to face discussion;
- Limitations of this study include the possible exclusion of “hard to get”, less participative and/or educated members of the CDG community as well as the fact that all think tanks were done in English;
- Potential difficulties of the implementation of the measures suggested by the participants include ethical, legal and social constraints associated with data exchange, ownership and donation of biological material;
- To overcome these potential obstacles a common dialogue space with all stakeholders needs to be implemented, so that common ground and group decisions can be made respecting the views of all people affected by CDG.

- The involvement and engagement of patients, families and professionals in health-related decisions is of particular importance in rare diseases' communities, as the needs and preferences of these communities are very often overlooked and not met efficiently;
- This approach can stimulate needs-driven research, which can more effectively respond to the pressing needs of a community, therefore having a fast and lasting impact on the quality of life of the community;
- A total of 48 participants were enrolled in this study and divided into 3 groups, being each group assigned to a think tank;
- Most participants had had contact with CDG for less than ten years (34/48) and half were involved in patient organizations.
- All think tanks were done in English;
- A semi-structured interview guide was used in all think tanks. Two main questions were asked:
  - 1) what are the challenges experienced by people living with or caring for patients with CDG?
  - 2) what strategies can be used to overcome those challenges?
- The Think Tanks were recorded, subsequently transcribed using strategies that assured the quality and rigour of the information;
- Participants felt that the complexity and lack of knowledge about CDG created many obstacles for the patients and families, difficult access to reliable information, quality health care and psychosocial support, and limited representation of CDG patients and caregivers in research and social matters;

<b>Unmet needs</b>	<b>Innovative solutions</b>
<u>Information</u>	<u>Civil society</u>
Difficult access to reliable and understandable information (particularly about experimental treatment);	Face-to-face and online peer support (there is an international private Facebook group called CDG Global Alliance )
Lack of doctors and medical professionals with expertise on CDG;	Mobilise more CDG families into advocacy to improve and extend lobbying and representation
<u>Health care</u>	<u>Care</u>
Diagnosis – usually a very complex process, with many delays and misdiagnosis;	Increase professionals' awareness, namely by including CDG in medical examinations, disseminating information on CDG, and adding CDG to diagnostics checklists;
Countries with poor access to genetic testing have very few CDG cases;	Improving care for CDG – develop specific quality care guidelines for CDG;
CDG are complex, multi-systemic diseases, which also makes it hard for healthcare professionals to develop an holistic approach to treatment and management of the patients;	Involve patients and families in decision-making, namely concerning treatment. For that patients need to be empowered and given information that allow them to make decisions;
Ultra-rare CDG forms are even more difficult to address, as clinical knowledge is more limited;	
Research on CDG has not been given enough funding;	<u>Digital</u>
Professionals are still resistant to collaborations;	Creation of an international, multilingual, continuously and stably funded online platform with updated information on CDG

Families lose trust on medical professionals	Patient registry with data and biological samples for research
<b>Psychosocial</b>	
Uncertainty about disease progression and prognosis;	
Lack of targeted treatments;	
Costly therapies and feelings of isolation;	
Necessity of full time caregivers, which usually leads to one of the parents quitting her/his job with also financial consequences	
<b>Representation</b>	
Only few families and patients are actively engaged in advocacy, leading to a misrepresentation of the community	

- Potential implementation hurdles of the measures suggested by the participants include ethical, legal and social issues regarding data exchange, ownership and donation of biological material;
- To ensure a smooth implementation of the ideas/solutions suggested, a common dialogue platform need to be built, in order to allow group and representative decisions to be made. Moreover, in this study only 1 adult CDG patient participated. The involvement of CDG patients is a challenge as most patients have speech difficulties. To make sure more CDG patients can be involved in future studies and express their views, other study designs need to be developed, one option being the use of creative visual methods (e.g. photography, video, acting performance)