WORLD CDG MAGAZINE

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> "I alone cannot change the world, but I can cast a stone across the waters to create many ripples."
>
> by Mother Teresa

"Keep in mind, you are not alone. Though Congenital Disorders of Glycosylation (CDG) are categorized as "rare" there is an amazing community at your fingertips, working day and night to improve the lives of many people living with CDG and their family members. We want you to know everything available for you in lay language, but we suggest you enter into it at your own pace and comfort level." By Vanessa Ferreira.

©World CDG Magazine, created and written by Vanessa Ferreira, Senior people-centric researcher and co-founder of the international research network CDG and Allies, Founder of the Portuguese Association for CDG (APCDG), Global Patient Advocate, and sibling to Princess Liliana.

Boost CDG Research, Education, and Awareness!

We rely on donations in order to continue our projects. Thus, we know how to do a lot with very modest budgets. However, we still need your donation to help us do our work. Support us

CDG and Allies is a people-centric international research network dedicated to CDG, based at FCT, NOVA University. WCDGO is led and operated by CDG & Allies Professionals and Patient Associations International Research Network (CDG & Allies PPAIN). CDG & Allies PPAIN is based at NOVA School of Science and Technology (FCT NOVA. For more information visit FCT NOVA). Discover everything about World CDg organization

CDG & Allies – PPAIN
CDG & Allies - Professionals and Patient Associations International Network

World Congenital Disorders of Glycosylation Organization (WCDGO) is the unified voice of people living with Congenital Disorders of Glycosylation (CDG). WCDGO is led and operated on a volunteer basis by several CDG & Allies research collaborators.

WORLD CDG ORGANIZATION
Awareness, Research, Education and Empowerment United
FAMILIES AND PROFESSIONALS

Our goals are:

1. Helping to find, organize, and empower people living with CDG and their families with an interest in forming new CDG patient groups or representing CDG in their countries;
2. Supporting our existing CDG patient organizations in their ongoing efforts to grow their organizations and raise CDG awareness, increase diagnosis and advocate for patient access to and reimbursement for modern life saving therapies;
3. Centralizing reliable information, resources, and initiatives aimed at educating people with CDG and boosting improved care and medical treatment.

We are dedicated to ensuring that no matter where families live, everyone diagnosed with CDG should benefit from good standards of care and trustable information worldwide.

World CDG Organization (WCDGO) mission is to provide a solid and unified voice to improve the lives of all people affected by CDG and their family members.

World CDG Organization (WCDGO) vision is to achieve the hopes of people living with CDG and their family members.

"The greatness of a community is most accurately measured by the compassionate actions of its members"

by Coretta Scott King.

Subscription

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Disclaimer

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Accordingly, we encourage you to consult with the appropriate professionals before taking any actions based on such information. We do not provide any medical or health advice. The use or reliance of any information contained in this magazine is solely at your own risk.

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Dear CDG friends,

Welcome to the first edition of our World CDG Magazine, another resource that provides a truly comprehensive overview of what is happening in the global CDG community

I would like to start this message by highlighting that the 6th World Conference on CDG, will be held in-person in Caparica, Portugal, at NOVA School of Science and Technology, FCT NOVA. **The 6th World Conference on CDG includes novelties aimed at securing global participation, thanks to 3 educational tracks:**

- The "World Think Metabolic, Think CDG Academy", composed of virtual pre-conference workshops delivered in english. It is intended for people living with CDG, their family members, academia, medical doctors, and the industry. It aims to provide CDG stakeholders with the knowledge and skills needed to become CDG research and drug development experts. It will be of great help to navigate along the overall sessions that will be delivered during the 6th World Conference on CDG.
- The "World CDG Advocacy and Leadership Academy" offers in-person sessions in Caparica, Portugal, at NOVA School of Science and Technology, FCT NOVA. It is tailored for leaders of CDG patient groups and their advocates and helps our CDG Organizations and representatives to be successful advocates.
- And the "6th World Conference on CDG" offers in-person sessions in Caparica, Portugal, at NOVA School of Science and Technology, FCT NOVA. It is intended for all CDG community. As simultaneous translation comes with a high additional cost, CDG and Allies will evaluate whispering translation if around a minimum number of people share the same language. Registrants cannot expect their preferred language to be added until CDG and Allies officially announce this.

More details about these events offerings can be found in this magazine or WorldCDG.org
As outlined in this magazine, we celebrate and share what can only be described as giants steps lead by research networks and industry stakeholders.

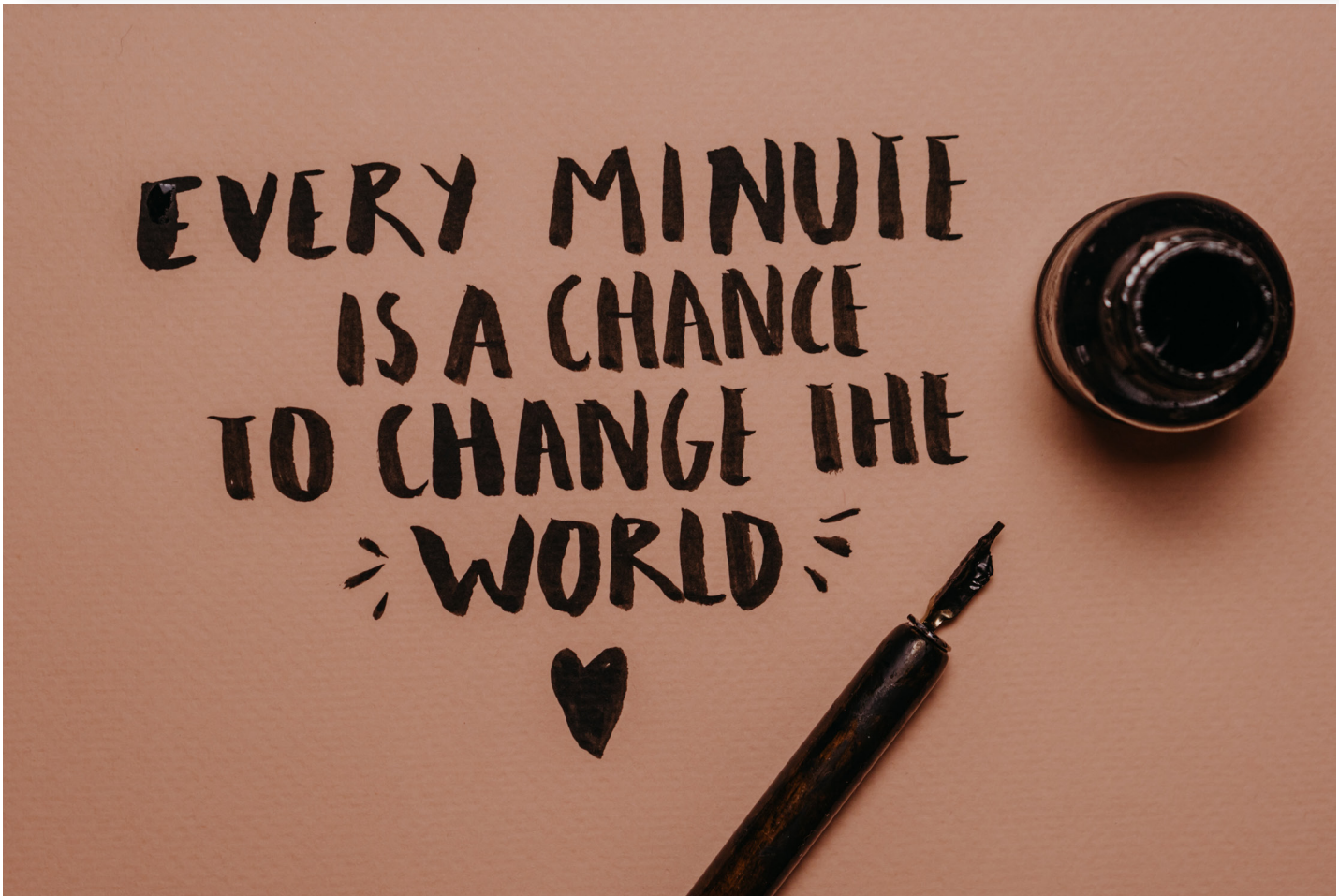
I wish you good health, happiness, and pleasant reading!

Warmest regards,

Vanessa Ferreira, PhD, MBA
Senior people-centric researcher and co-founder of the international research network CDG and Allies, Founder of the Portuguese Association for CDG (APCDG), Global Patient Advocate, and sibling to Princess Liliana.

> It is of extreme importance that every person in the CDG community becomes one united voice and takes every conquest in a specific CDG as a victory for the whole community. It is important to keep this in mind because CDG are all connected, and a new finding regarding one of them will give tools to improve basic and therapeutic research in the others.
>
> By Vanessa Ferreira, sister to Princess Liliana, who lives with CDG.

How can you help our CDG community? Is easy! Share the information we make available within this magazine and World CDG Organization website among your social media and other channels. This raises awareness, accelerates diagnosis, and secures better care and management for our CDG children and adults!



> “Find a group of people who challenge and inspire you, spend a lot of time with them, and it will change your life”
>
> by Amy Poehler

Love is the common language within CDG Patient groups and advocates. Finding a powerful CDG community is just a click away. Go here.

ADVOCATING AND HELPING CDG FAMILIES IN GEORGIA

BY TATA TSINTSADZE, FROM CDG GEORGIA!

Our beautiful Tata, mom to Natalia from Georgia.

I am Tata Tsintsadze, 37, from Tbilisi, Georgia. I have a beautiful daughter Natalia who has PMM2-CDG. I am absolutely grateful for having two incredible daughters, my family and friends, my job and things I like and enjoy doing in my life.

One of the biggest joys in my life is to be able to help people and see their happy faces.

The quote I tell to myself every single day is "Do one kind deed every day!". If people were more kind the world would be a better place.

Georgian national music and songs is what gives me energy, inspiration and good mood.

Today my daughter Natalie was able to walk on her own, and better than yesterday. Her face was shining!

"Do one kind deed every day!"

The story of Tata and Natalia is also found here. I would like to help Georgian CDG families and patients within my organization 'CDG Georgia', to be able to support them financially as well as inspiring them emotionally, so that our children have a brighter future, equal integration in the society and better health.

Learn more about Tata Tsintsadze, Founder of CDG Georgia here.

You can reach out to Tata here. Follow CDG Georgia on Facebook here.

Our wonderful Natalia, from Georgia.

Patient driven CDG Research initiatives are underway!

By Andrea Miller, from CDG CARE!

As part of CDG CARE's effort to advance and diversify patient-led research in the fields of CDG and NGLY1-CDDG, CDG CARE now offers fiscal sponsorship opportunities for well-defined and collaborative research projects. Through this program, CDG CARE sponsors new research and time-limited projects, as well as more established grassroots projects that are mutually agreeable and advance both the project and CDG CARE's mission.

Over the past few months, CDG CARE is delighted to share that research programs have been launched in several areas including GMPPA-CDG, PIGA-CDG, PIGN-CDG, PIGS-CDG, and ALG8-CDG! Through these partnerships with families, over $350,000 has been raised in just a few months' time!

To learn more about the benefits that a partnership with CDG CARE can provide, please review CDG CARE's new Fiscal Sponsorship Program by Clicking here.

Our beautiful Andrea Miller, mom to Bianca and President to CDG CARE.

To learn more about the projects that are currently underway, simply click on the links below:

Bloom for a cure – Advancing Research for GMPPA-CDG
Drug Repurposing Project for PIGA-CDG New Treatment
Finding a Treatment for PIGN-CDG
Help Find a Treatment for PIGS-CDG
Project GRACe – Glial Research in ALG8-CDG

You can reach out CDG CARE here. Follow CDG CARE on Facebook here. And Instagram here.

News from CDG Ambassadors

CDG Ambassadors represent a CDG for which in many cases there is not yet at country level a legal non-profit organization. Our CDG Ambassadors wish to connect with other families, to enable support, to exchange experiences, to create critical mass and to raise awareness and education for a certain CDG. They constantly strive to improve the situation for many CDG families over the globe – no matter how they are organized. **Let's meet some of them!**

SANJA JURIC, CROATIA AND GERMANY

COG4–CDG AMBASSADOR

"After hearing the diagnosis, which we never knew before, we felt like we were alone in the world."

There is one home surrounded by flowers, a source of warmth and love, where lives a small family. Dad Srećko, the best dad in the world, mom Sanja and greatest gift that God can give, our three-year-old daughter Mila, our smiling fairy.

At seven months, Mila was diagnosed with COG4-CDG syndrome. After hearing the diagnosis, which we never knew before, we felt like we were alone in the world.
And then our fight begins, our real life. Most significant of all was finding a group of parents who write about CDG. Then I finally got the feeling that we belong somewhere, that people somewhere understand us. I can only say a big thank you to Vanessa for all your hard work. The influence of the World CDG Organization on our lives is really meaningful.

I can't say at what point we accepted the diagnosis; it happened over time. No matter how difficult it was, we learned that Mila was not her diagnosis. She is a small joyful girl who wants to know the world, who feels and loves the world, who has enriched not only our lives, but many other lives as well.

Our beautiful Mila and her amazing mother!

At the end of the day, I am grateful because I have Mila, just the way she is. I am grateful because I am her mother. A mom who hasn't even heard the word mom today. But today I am the proudest and best mom in the world, because I feel that pure soul hidden under its limitations. And when she is the quietest, I hear her thoughts hidden in this ordinary world. I am grateful for her diagnosis because without her I might never have discovered the secret of life. And that is living today. Stay in a moment, the birds are singing so beautifully, do you hear them?

I am grateful for wonderful family and friends, whose support is immeasurable. I am grateful because my soul and my vision have become clearer. And now I can stop and absorb such precious moments. If I happen to forget it and gloomy clouds hang over me, Mila reminds me. And it's so incredible, as if she senses that I need strength to continue.

"I want to tell parents, don't be afraid! After finding out the diagnosis of the most loved child is not easy. Cry it out, take your time, and slowly move forward. And know that you are not alone in the world."

Our every day is filled with music, from classics, religious songs that we really love to rock n' roll, with which we make a party. We turn on the disco ball, turn up the music and we sing and dance until our feet hurt. And Mila is smiling like the happiest child in the world. And I truly believe that she is the happiest child in the world. There are many moments like this, when my heart sings while I watch her full of joy.

That's why I want to be her voice, so that the world hears her and that the diagnosis of COG4-CDG means a happy and blessed life. A life filled with laughter and joys that you can't even imagine. A life that is not a fairy tale and that is not at all easy, but a life that gives so much.

I want to tell parents, don't be afraid! After finding out the diagnosis of the most loved child is not easy. Cry it out, take your time, and slowly move forward. And know that you are not alone in the world. You will laugh, discover joy where you least expect it and it will be immeasurable. You'll be fine, trust me, I've been where you are now.

"Start with what is necessary, then do what is possible and you will quickly do the impossible"

St. Francis of Assisi.

Read more about COG4-CDG

WANDA KADZIOLKA, CANADA
SSR4–CDG AMBASSADOR

Our incredible Wanda Kadziolka, SSR4-CDG Ambassador.

My name is Wanda Kadziolka and I am the mother of Andrew Kadziolka, born July 12 1995. He was diagnosed with SSR4-CDG in 2019. He represents our SSR4 group of 10 children of different ages.

Andrew is 27 years old now. Currently, he is the eldest child with SSR4 that we know of. We live in Ottawa, Canada.

Andrew's birth completely changed our family life. We had to adjust to a new situation. Our family members' needs were put on the back burner. Almost overnight, from a professional woman, I turned into a stay at home mom. The worst thing was that we did not know why Andrew suffered so much and how to help him. We went through a nightmare.

I am grateful that after Andrew was finally diagnosed in 2019, I found information about CDG Facebook and I was welcomed to join. I feel that I am not alone and I have support I appreciate very much. Big thanks to Vanessa and Andrea for their dedication, time and hard work.

I am grateful for CDG conferences because we learn so much from meeting specialists and sharing information with doctors, scientists and parents. Attending the conference in San Diego in 2020 was a starting point for me. I met wonderful people, learned about CDG syndromes and made plans for a future. Now Andrew is under the care of an exceptional doctor, and I am very grateful for that.

I am grateful for my daughter, Marta's support, research and encouragement. She is constantly looking for new possibilities to improve her brother's life.

I am grateful for my husband's support and time even though I do not have much time for him because of Andrew's needs. And I am very grateful to our Lord for the strength He gives me.

> "My daily inspiration is to make every day count for Andrew. To do my best!"

My heart sings when I hear my Andrew laugh and smile and when he hugs me.

My heart sings when I hear about new CDG research and hope that it can improve Andrew's life.

My daily inspiration is to make every day count for Andrew. To do my best!

A lot of things brighten my day: Andrew's laugh, music, audiobooks (no time to read).
I like Yanni, Edith Piaf, Jean Michel Jarre, Bach. Bach's Air calms me down. I always enjoy listening to Yanni, Edith Piaf encourages me as well as the tango scene from Scent of a Woman to dare to try new things. Latin music puts fire in my blood. I like to listen to books by Bernard Cornwell, Ken Follet. Rhys Bowen books always relax me if I am having a difficult day.

What made my day today? Andrew had dental surgery. Except for a few "bumps" on the road, everything went well. He went under general anaesthetic. Now Andrew's teeth look great, he smiles beautifully and his dentist was extremely kind and did a super job!

Advocating for CDG in general and especially for SSR4 means to me spreading knowledge about the disorder, sharing information, keeping it in our mind always and reaching for possibilities to advocate whenever we can. I am reaching out to Ottawa politicians and I have found SSR4 infographics very helpful to inform about disease.
I am personally ready to share my experience with SSR4 obtained through 27 years of Andrew's life: Andrew's problems, solutions, medication and treatment. My daughter and I established the CDG-1y-SSR4 group in order to keep SSR4 together and make it easier to share information, solutions and talk to each other, socialize.
We are still working on how to make it better and we have a lot to learn from Vanessa and Andrea.

Learn more about SSR4-CDG, view or download the infographic available at WCDG.org:

SSR4-CDG Infographic

Infographic

Disorders (CDG Types): SSR4-CDG

Year: 2021

Authors: Marta Falcão, Carlota Pascoal, Andrew, Wanda and Marta Kadziolka, Tatiana Soffiati, Andrew Edmondson, Vanessa Dos Reis Ferreira, Prof. Jaak Jaeken

Languages: English

Keywords: CDG, Congenital Disorders of Glycosylation, SSR4-CDG

View/Download

We need you!

Do you want to boost awareness and representativeness for a specific CDG type?

Write us until February 8, 2023:

https://worldcdg.org/contact

Designed by Vectpezy

The community-centric international research network, CDG & Allies, as well as the World CDG Organization, want to ensure that ALL CDG types are represented in everything we do. We have many activities planned aimed at #RaisingAwareness and #Education about different CDG types.

We're counting on you! Write us

News from CDG & Allies and World CDG Organization Collaborators

Let's welcome Marisa Godinho!

We are happy to announce the recent arrival of one more member of our team, Marisa Godinho!
Marisa will be joining us to help CDG and Allies and World CDG ORG activities.
Marisa, thank you for taking on this challenge and for joining this family!
With Marisa, the future looks bright, don't you think?

As good news is never too much.... Let's welcome Ana Ferreira Verde!

Please join us in officially welcoming a new member of our team, Ana Verde! We are so happy and excited to see our team growing.
Ana will be joining us to help CDG and Allies and World CDG ORG activities. Check more here.
Ana, thank you for taking on this challenge! United we are stronger!

Let's welcome Pedro Granjo!

Pedro will be fully dedicated for CDG until July 2023! He will drive CDG research, advocacy, awareness and educational activities for our community!
The Liliana's Scholarships are funded by the Portuguese Association for CDG and the CDG and Allies international network.
Pedro, Thanks for all help you gave so far as a volunteer! We all look forward for your next steps!

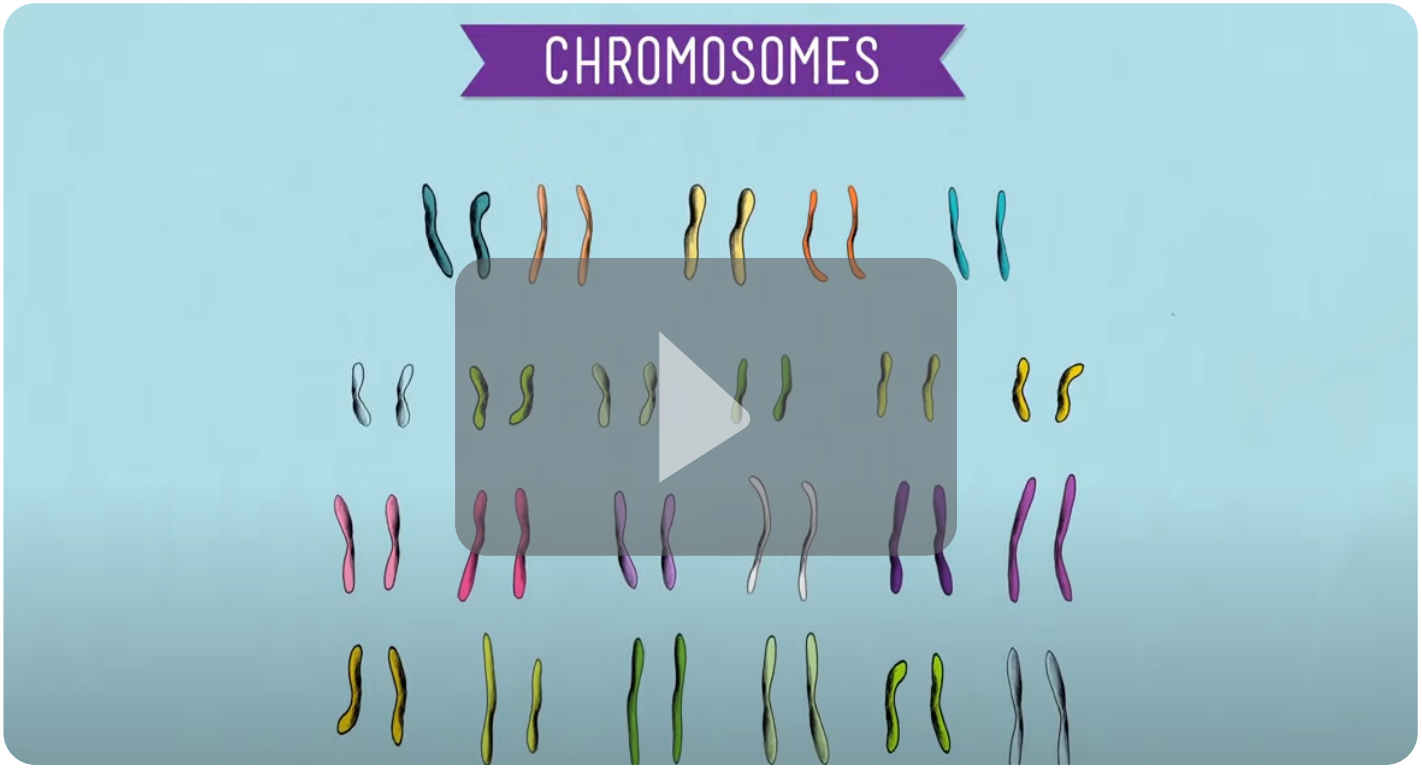
What's new for me to learn and get empowered?

What is CDG?

We want to empower our community! A new section dedicated to What is CDG was launched! Check here

What causes CDG?

Learn more about key concepts in CDG world. This can help you, explain easily what CDG is, the next time you are with your family, friends or doctors.
First, let's read here at what causes CDG. Check out the following video below where Hank and his brother John discuss heredity:



Continue learning here.
*The WorldCDG.org website gathers information about therapies for CDG from reliable databases to make it easier for patients, families, and caregivers to find the information they seek. Also note that due to advances in science, knowledge grows very fast. Do not hesitate to contact us

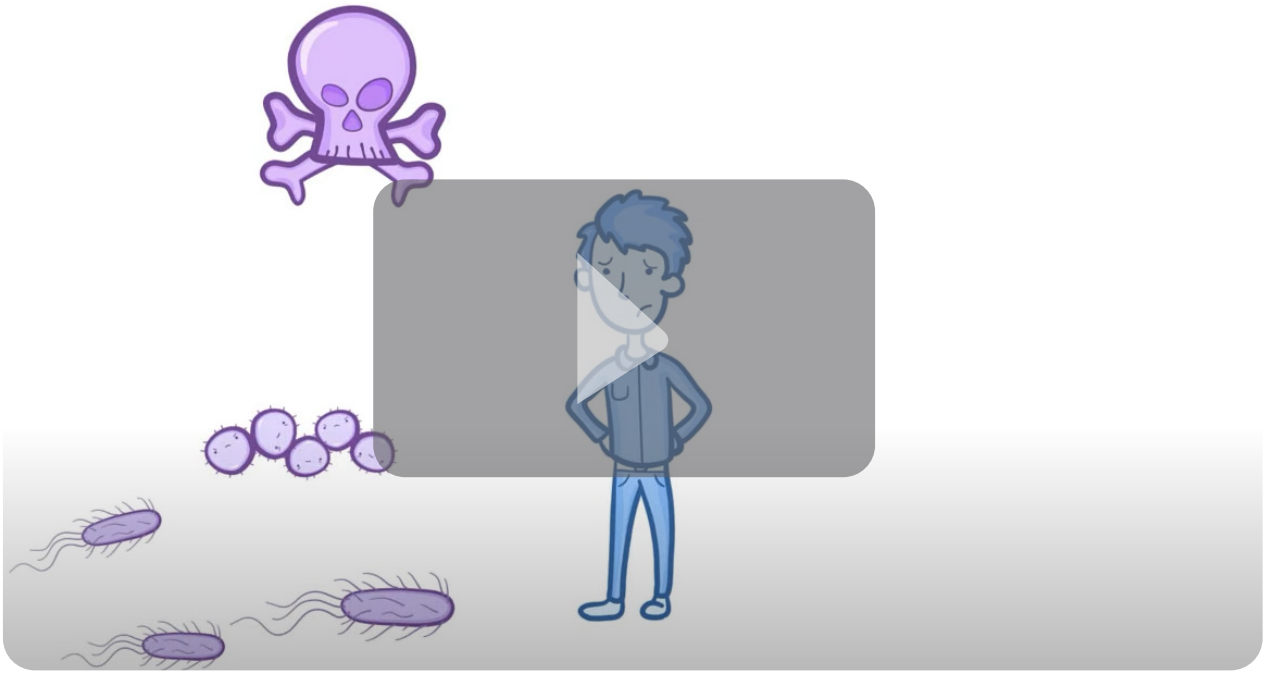
New content about CDG Immunology:

Roll the drums... A new section about #Immunology is now available on our website!

Many materials, such as videos, articles, and images, were created with and for our #CDGCommunity to better understand how the immune system works in CDG.

What is the immune system and why is it important?

The immune system consists of a network of special cells, tissues, and organs that protect the body. Osmosis offers a video with an introduction to the immune system:



To understand how important, it is in CDG read more here.
*The WorldCDG.org website gathers information about therapies for CDG from reliable databases to make it easier for patients, families, and caregivers to find the information they seek. Also note that due to advances in science, knowledge grows very fast. Do not hesitate to contact us

What is Quality of Life?

The World Health Organization (WHO) defines Quality of Life (QoL) as an individual's perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns.

QoL is a concept that depends on multiple aspects of people's lives:

Source: https://ec.europa.eu/eurostat/web/quality-of-life/data

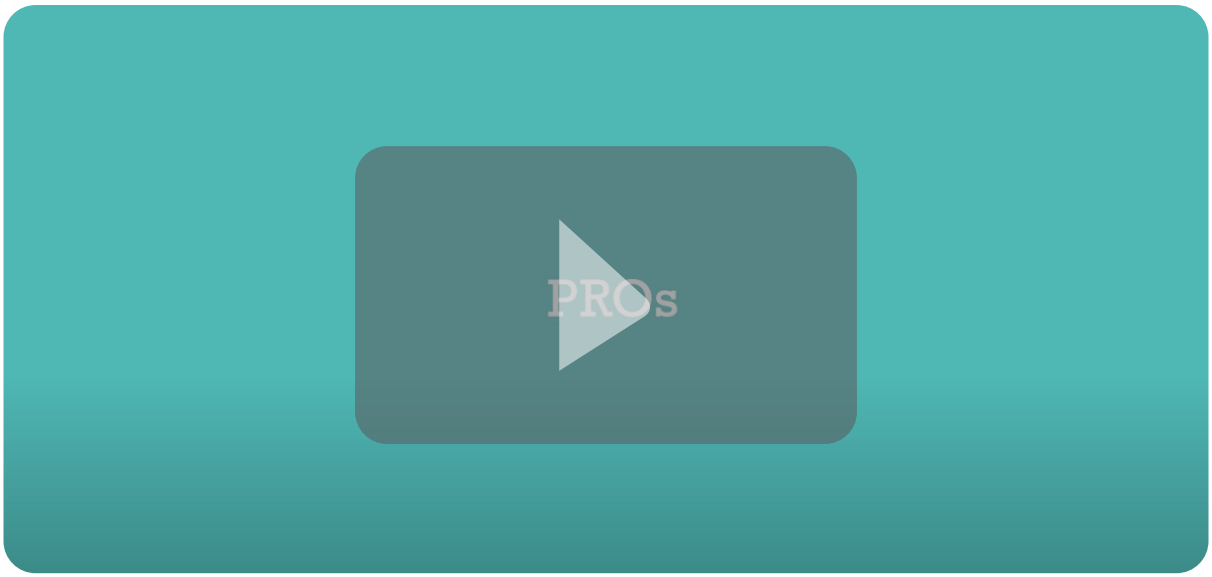
What is health-related quality of life (HrQoL) and why is it important?

Health related quality of life (HrQoL) is the health aspect of QoL that focuses on people's level of ability, daily functioning and ability to experience a fulfilling life.
HrQoL research is pivotal for any population and/or disease. It helps people understand how health, disease and treatment impact the QoL.
People are then better able to, for example :

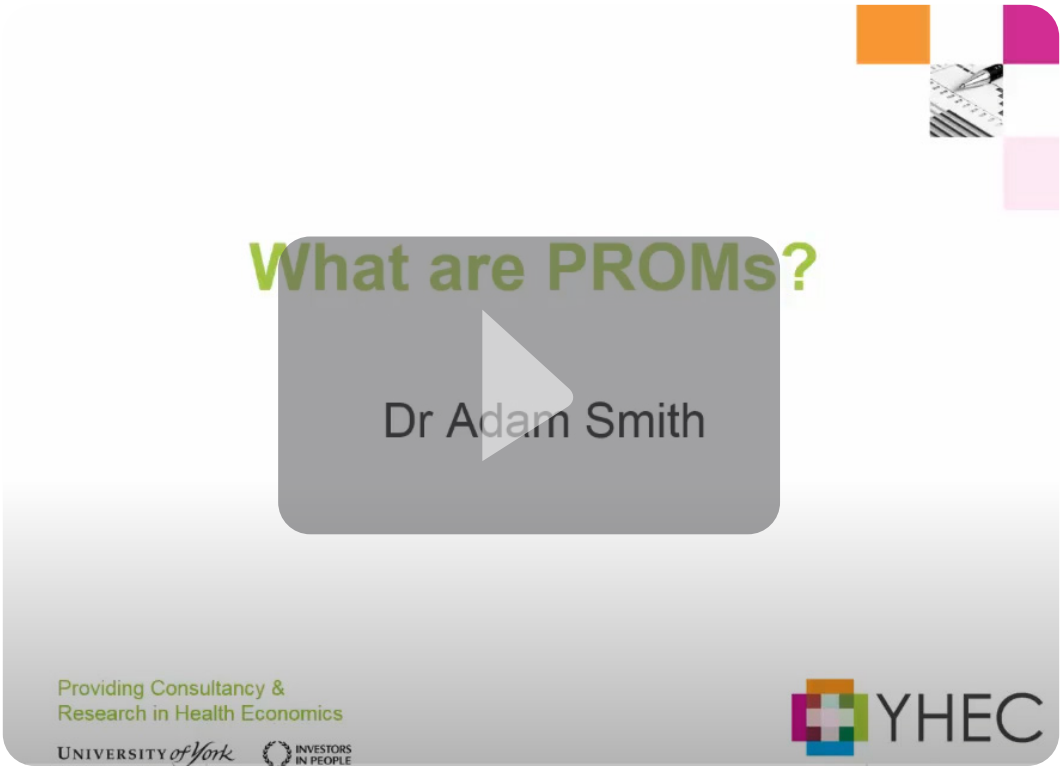
1. Understand the potential benefits and risks of a proposed treatment
2. Weigh the impact of a decision on symptoms, function and life expectancy
3. Live their lives more fully

What are PROs, PROMs and ObsROMs?

- **PROs** - **Patient reported outcomes** - are any report of the status of a patient's health condition or health behaviour that comes directly from the patient, without interpretation of the patient's response by a clinician or anyone else. Self-reported patient data provide a rich data source for outcomes. Learn more at WorldCDG.org. To better understand what PROs are, watch this video from the Canadian Partnership Against Cancer:



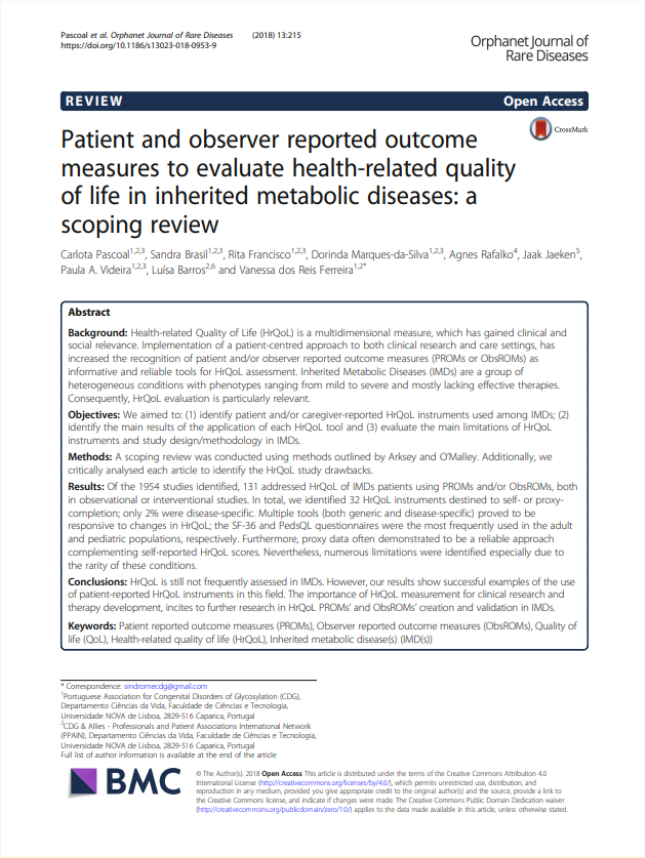
- **PROMs** - **Patient reported outcome measures** - PROMs are tools used to collect patient-reported outcomes directly collected from patients/people living with a disease. Learn more at WorldCDG.org. To better understand what PROMs are, watch this video from Dr. Adam Smith:



- **ObsROMs** - **Observer-reported outcome measures** - tools that allow to obtain reports made by a proxy (a person who is in direct contact with the patient) when it is not possible to obtain self-reports (e.g. in case of intellectual disability). Learn more at WorldCDG.org.

HrQoL and PROMs/ObsROMs and in rare diseases and in CDG

While some rare diseases do not necessarily affect life expectancy, the majority lead to physical, emotional and/or psychosocial limitations with a wide range of disabilities. This can be due to their chronic nature, complexity, severity and even the lack of treatments. Read more at WorldCDG.org.



Pascoal *et al. Orphanet Journal of Rare Diseases* (2018) 13:215
https://doi.org/10.1186/s13023-018-0953-9

Orphanet Journal of Rare Diseases

REVIEW — Open Access

Patient and observer reported outcome measures to evaluate health-related quality of life in inherited metabolic diseases: a scoping review

Carlota Pascoal[1,2,3], Sandra Brasil[1,2,3], Rita Francisco[1,2,3], Dorinda Marques-da-Silva[1,2,3], Agnes Rafalko[4], Jaak Jaeken[5], Paula A. Videira[1,2,3], Luísa Barros[2,6] and Vanessa dos Reis Ferreira[1,2*]

Abstract

Background: Health-related Quality of Life (HrQoL) is a multidimensional measure, which has gained clinical and social relevance. Implementation of a patient-centred approach to both clinical research and care settings, has increased the recognition of patient and/or observer reported outcome measures (PROMs or ObsROMs) as informative and reliable tools for HrQoL assessment. Inherited Metabolic Diseases (IMDs) are a group of heterogeneous conditions with phenotypes ranging from mild to severe and mostly lacking effective therapies. Consequently, HrQoL evaluation is particularly relevant.

Objectives: We aimed to: (1) identify patient and/or caregiver-reported HrQoL instruments used among IMDs; (2) identify the main results of the application of each HrQoL tool and (3) evaluate the main limitations of HrQoL instruments and study design/methodology in IMDs.

Methods: A scoping review was conducted using methods outlined by Arksey and O'Malley. Additionally, we critically analysed each article to identify the HrQoL study drawbacks.

Results: Of the 1954 studies identified, 131 addressed HrQoL of IMDs patients using PROMs and/or ObsROMs, both in observational or interventional studies. In total, we identified 32 HrQoL instruments destined to self- or proxy-completion; only 2% were disease-specific. Multiple tools (both generic and disease-specific) proved to be responsive to changes in HrQoL; the SF-36 and PedsQL questionnaires were the most frequently used in the adult and pediatric populations, respectively. Furthermore, proxy data often demonstrated to be a reliable approach complementing self-reported HrQoL scores. Nevertheless, numerous limitations were identified especially due to the rarity of these conditions.

Conclusions: HrQoL is still not frequently assessed in IMDs. However, our results show successful examples of the use of patient-reported HrQoL instruments in this field. The importance of HrQoL measurement for clinical research and therapy development, incites to further research in HrQoL PROMs' and ObsROMs' creation and validation in IMDs.

Keywords: Patient reported outcome measures (PROMs), Observer reported outcome measures (ObsROMs), Quality of life (QoL), Health-related quality of life (HrQoL), Inherited metabolic disease(s) (IMD(s))

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Full list of author information is available at the end of the article

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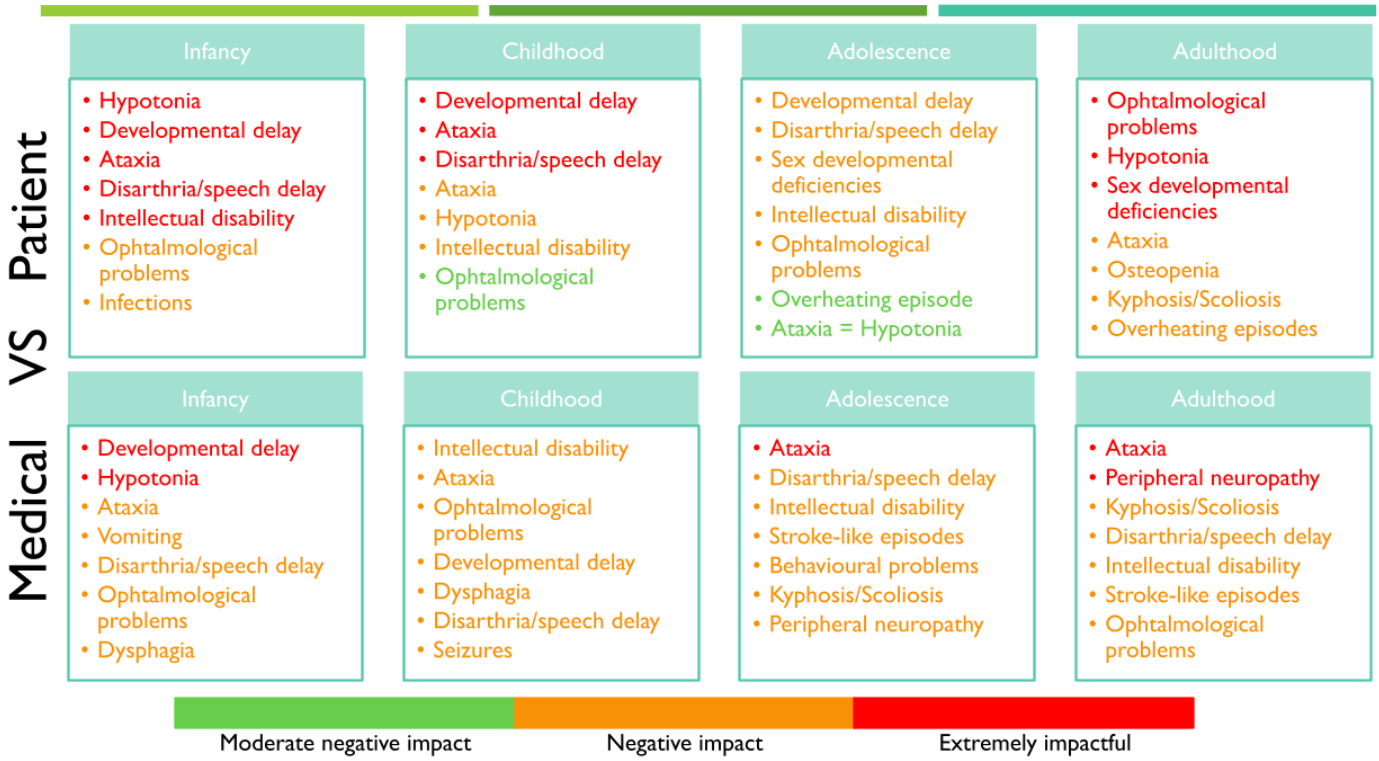
Current research dedicated to QoL in CDG

Given the current state and importance of QoL knowledge in the CDG field, the CDG & Allies international research network has been making significant contributions.
Back in 2018, the CDG & Allies team reviewed the literature and gathered the PROMs and ObsROMs that are used in other inherited metabolic disorders, like CDG. Despite the CDG particularity, some aspects of these diseases are similar to the ones of CDG and therefore, some key learnings were identified for future research.
To access the full article, please click HERE.

However, CDG complexity and heterogeneity makes them a unique set of diseases. The severity and burden between patients and their families can be rather disparate. Consequently, there is no one-size fits all solution that will cover the needs for all the families affected by these conditions. Particularly in this scenario, it is particularly important to hear the voice of the community, and be as inclusive as possible and value the needs of each individual.
That is why CDG & Allies are working towards a creative and tailored solution for the CDG community, promoting the engagement of all expert stakeholders, including CDG families, medical professionals, and pharma representatives.

Focusing firstly on PMM2-CDG, an expert committee was created to identify what aspects of the disease constitute a major burden for the patient's quality of life. Importantly the vision of the medical doctors and patient caregivers complemented each other. On the one hand, caregivers highlighted aspects related to the real-world daily living which might not be visible during medical appointments. On the other hand, medical doctors can contribute with their extensive knowledge and experience derived from dealing with many different patients during many years.

A sum up of the main CDG impactful signs and symptoms across the patient lifetime can be found below:



Continue learning more about this relevant topic for CDG at WorldCDG.org

*The WorldCDG.org website gathers information about therapies for CDG from reliable databases to make it easier for patients, families, and caregivers to find the information they seek. Also note that due to advances in science, knowledge grows very fast. Do not hesitate to contact us.

Epidemiology in CDG

Did you know that...In our website there is a new section dedicated to Epidemiology?

What is Epidemiology and why is it important?

The word "epidemiology" comes from the Greek words epi, meaning on or upon, demos, meaning people, and logos, meaning the study of. In other words, the word “epidemiology” has its roots in the study of what befalls a population. Many definitions have been proposed, but the following definition captures the underlying principles and public health spirit of epidemiology:
Epidemiology is the study of the distribution and determinants of health-related states or events in specified populations, and the application of this study to the control of health problems (1). To continue reading about the definition of epidemiology visit the book developed by the Centers for Disease Control and Prevention (CDC) available here.

Let's Learn Public Health prepared a video, where an overview of the most common types of epidemiological studies, their advantages and disadvantages is given. It also looks at systematic reviews and meta-analysis. Watch it below:

Learn all about it at WorldCDG.org.

*The WorldCDG.org website gathers information about therapies for CDG from reliable databases to make it easier for patients, families, and caregivers to find the information they seek. Also note that due to advances in science, knowledge grows very fast. Do not hesitate to contact us.

Therapies for CDG

What is drug repurposing?

Watch the introductory video by Findacure about drug repurposing, aimed at small rare disease patient groups.

Read and share about therapeutic approaches in CDG at WorldCDG.org

Everything you need to know in our new section at the World CDG Organization website!

*The WorldCDG.org website gathers information about therapies for CDG from reliable databases to make it easier for patients, families, and caregivers to find the information they seek. Also note that due to advances in science, knowledge grows very fast. Do not hesitate to contact us.

Peer-to-peer support: Spotlight to Ask Me Anything

What is Ask Me Anything (AMA)?

Ask Me Anything (AMA) sessions are opportunities for you to meet and interact with special CDG guests such as researchers, leaders, innovators, entrepreneurs, and more. During these sessions, guests will answer live any questions you have about their work, areas of expertise, and anything in between. The Ask Me Anything series allows you to increase your knowledge about CDG. The series also provides an opportunity for you to know that you aren't alone! The objective of shining this light is to continually increase the support to our community and move forward toward a culture of inclusivity and a sense of belonging.

Who can attend? All sessions are intended for people living with CDG, family members, and caregivers.

How is Ask Me Anything held? Ask Me Anything will be held in the form of a virtual webinar using the Zoom Application. The host is in charge of facilitating the technology used. The moderator or session leader coordinates each session and facilitates the question-and-answer session.

All meeting Ids, passwords, and technical meeting schedules are sent to attendees' emails. If you have questions, write us.

Who is our special CDG guest? Pf Jaak Jaeken accepted this challenge. And will be with us during the 3 first Ask Me Anything.

When is taking place Ask Me Anything? Check the calendar here.

Image: Last November, we launched the first Ask Me Anything. We were privileged to count with Pf Jaeken as special guest!

Meet prominent researchers, leaders, innovators & more, and ask them anything!

Register now here until 13 January 2023 for the next Ask Me Anything!

Awareness

New website section dedicated to International Awareness days and months

What is an International Awareness Day?

Each international day gives many people the chance to plan events related to the day's theme. An international day is used by organizations and offices of governments, civil society, the public and private sectors, schools, universities, and, more generally, citizens to **keep raising awareness on a certain theme**.

Which are the international days that the World CDG Organization is joining?

International Awareness Days are opportunities to raise awareness about important global concerns, generate support and funding to address certain challenges, and celebrate and reinforce human progress. In order to raise awareness for CDG, the World CDG Organization is joining several days. Please check them all here or within our sections dedicated to events here.

Please note: Some organizations extend their awareness activities for the entire month instead of one single day. Like that, they amplify their voices and raise more awareness. This is the case for **Rare Disease Day**, which falls on the last day of February but is celebrated throughout the whole month, or even **Health Literacy Day**, which falls on September 8, but is usually celebrated throughout the whole month of October.

New website section at WorldCDG.Org dedicated to Rare Disease Day

What is Rare Disease Day?

Rare Disease Day is held every year on February 28 (or February 29 in leap years), which is the rarest day of the year. It is a globally coordinated movement on rare diseases that works for equity and fairness in social opportunities, healthcare, and access to diagnosis and treatments for people living with a rare disease.
Since its creation, Rare Disease Day has played a critical role in building an international rare disease community that is multidisciplinary, global, diverse, and united with a purpose - Raising awareness and generating change for the people around the world who live with a rare disease, as well as their families and caregivers.
The purpose is to bring families with rare disease diagnoses together so they can build their network, talk to researchers, and learn more about the resources and services that are available to them.
For the 15th edition of Rare Disease Day in 2022, over 100 countries and regions have joined their voices, organizing over 600 different types of events around the world. Check the official video for the Rare Disease Day 2022 below:

Learn more in our new section

World CDG Day

MAY 16, WORLD CDG DAY

Is an annual event that provides international focus for patient groups and families living with CDG. It is an opportunity to raise awareness, influence change, promote timely disease diagnosis, and improve access to better Standards of Care and Management.

The Portuguese Association for CDG (APCDG), first launched World CDG Day in 2016 together with CDG CARE and worldwide CDG patient groups, advocates and professionals. Since that time, more than 300 events have taken place around the world, generating strong public and media interest.

The World CDG Day has also received support from governments worldwide, high-profile Non-Governmental Organizations (NGOs) and supranational bodies.

World CDG Day has laid the foundation for sustained global disease awareness and every year as the Campaign takes shape, a new theme is promoted to engage audiences in interactive ways, aiming to reach more people than ever before.

The #CDGMosaic is an important piece of art launched in 2021 that aims at honouring our CDG children and adults. It reminds us that we are united together in making a profound difference in the lives of people living with CDG.

Take a look at the free materials available in 2022 to our community to support World CDG Awareness Day

Let's get ready for World CDG Day 2023!

LET'S WALK THE PORTUGUESE CAMINO DE SANTIAGO TOGETHER?

The Camino de Santiago known in English as the Way of St James, is a network of pilgrims' ways or pilgrimages leading to the shrine of the apostle Saint James the Great in the cathedral of Santiago de Compostela in Galicia in northwestern Spain, where tradition holds that the remains of the apostle are buried.

Guess what? Vanessa Ferreira wishes to do part of the Camino to raise awareness for CDG. This is an event for everyone interested in helping to spread CDG awareness by walking together with Vanessa.

DO YOU WANT TO JOIN VANESSA?

Write us expressing your wish to walk "El Camino"

follow the link: https://worldcdg.org/index.php/contact

When	Route
8 May 2023	**Day 1:** Arrival to Caminha
9 May 2023	**Day 2:** Caminha – Vila Nova de Cerveira (15 km)
10 May 2023	**Day 3:** Vila Nova de Cerveira – Valença (15 km)
11 May 2023	**Day 4:** Valença – O Porriño (20 km)
12 May 2023	**Day 5:** O Porriño – Arcade (22 km)
13 May 2023	**Day 6:** Arcade – Pontevedra (13 km)
14 May 2023	**Day 7:** Pontevedra – Caldas de Reis (22 km)
15 May 2023	**Day 8:** Caldas de Reis – Padron (19 km)
16 May 2023	**Day 9:** Padron – Santiago (25 km)
17 May 2023	**Day 10:** Departure

DO YOU HAVE MORE IDEAS FOR OUR WORLD CDG DAY 2023?

SUBMIT IT NOW

Resources for our community

The CDG and Allies international research network, receives many requests for information about different types of CDG. Afterwards, develops summaries tailored for the medical community. Then, these are shortened and made available for our CDG community in a lay-language infographic format. The use of non-technical terms in order to share information in a simplified manner, is oriented towards a lay non-scientist audience who lacks the academical/technical background in the field. Check all infographics available here. These resources can easily be used to raise awareness for CDG. Contact us to learn more.
Shining light to DOLK-CDG, COG4-CDG and RFT1-CDG.

Events and trainings

Several events considered of interest to our CDG community are available at WorldCDG.org. They are organized per topics including:

- CDG
- Digital health
- Glycobiology
- Health literacy and lay language
- Health Technology Assessement
- Immunology
- Mental health and well-being
- Metabolic diseases
- Participatory medicine
- Patient engagement
- Patient experience
- Quality of Life (QoL)
- Rare diseases

A diverse range of trainings that can upskill our CDG community are available here

If you know of an event that is currently missing in our dedicated section, please e-mail us your information.

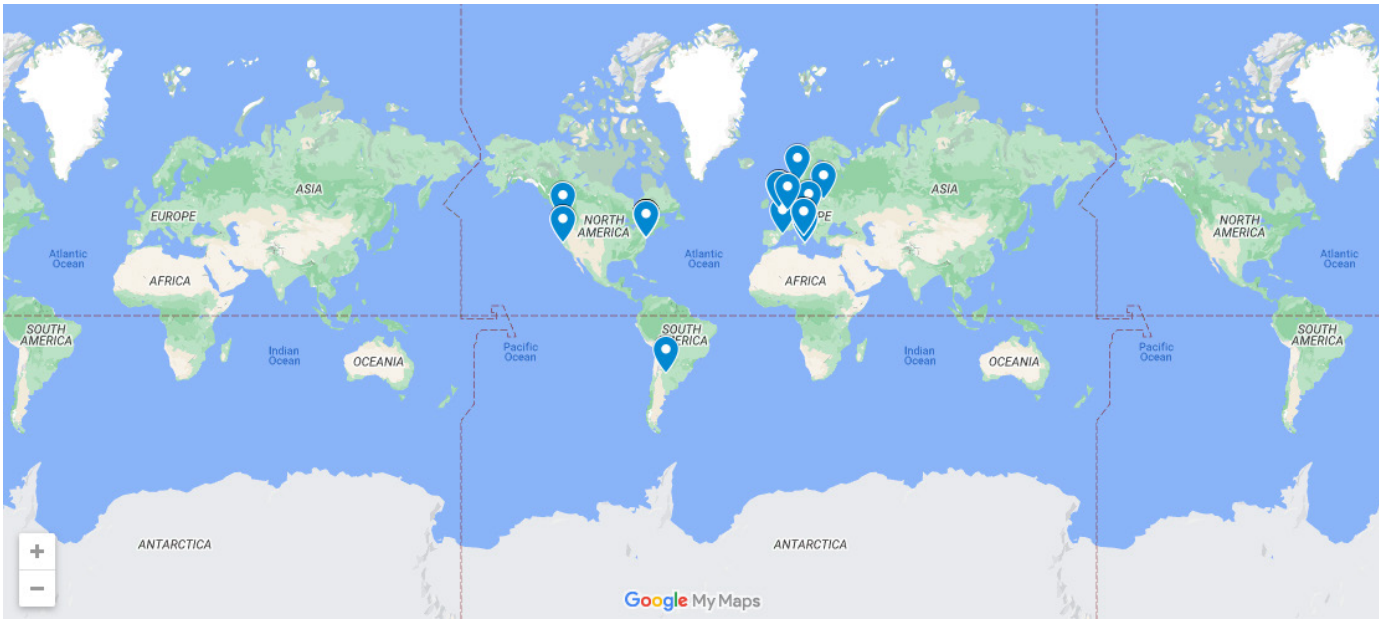
WorldCDG.Org launched a new section dedicated to CDG care and management!

Families with CDG often have a hard time finding diagnostic and research labs that know about it. In many parts of the world, people may never get the definitive diagnosis they desperately need to get the right care because they can't get to a lab or to a clinician who suspects CDG.

We are making a map of CDG by putting all of this information together, and you are welcome to tell us about labs that are leading the way in CDG diagnosis and research by filling out a survey form. Moreover, CDG patient groups can help point you in the right direction. Learn more here.

Would you like to feature your work in CDG or a reference care centre that leads care and management for people living with CDG in your country in our CDG Map?

It's easy! Complete the CDG Map Collecting Survey, found at the end of this webpage here!



Advocacy and Policy

EURORDIS represents the voice of patients and in this capacity plays a central role in the policy making for rare diseases.

- Learn more here about the important role **national policy** plays in improving the lives of people affected by rare diseases and what EURORDIS does to support the development of national rare disease plans.
- Learn more about EURORDIS' advocacy work to influence **European policy makers** including at the European Commission and European Parliament. Read here.
- Discover about the actions being taken at the **international level, at the UN and WHO, to make rare diseases a global health priority**. All information here.

About World Conference on CDG (WCCDG) 2023

This year's theme is **"Building a collaborative, community-centric strategy to boost drug development for all CDGs"**! and includes novelties like the **"World CDG Advocacy and Leadership Academy"** and the **"World Think Metabolic, Think CDG Academy"**! Stay tuned! Learn everything here.

The 6th World Conference on CDG, taking place in Caparica, Portugal, at NOVA School of Science and Technology, FCT NOVA, will be the largest, ever global gathering of People Living with CDG (PLwCDG), caregivers, healthcare professionals, and the pharmaceutical industry. It is the most complete and resourceful international conference focused entirely on CDG. The content is delivered in a patient-friendly language that non-specialist audiences can understand. The ultimate goal is to help people to engage with clinicians and researchers more effectively and participate actively in their healthcare decisions. Check the preliminary programme here. Key dates here.

The novelties include:

- **World Think Metabolic, Think CDG Academy**, May and June 2023, is composed of virtual pre-conference workshops. It is intended for people living with CDG, their family members, academia, medical doctors, and the industry. The virtual pre-conference workshops are delivered in English. The "World Think Metabolic, Think CDG Academy" aims to provide CDG stakeholders with the knowledge and skills needed to become CDG research and drug development experts. Thus, by securing your attendance and actively participating during the virtual pre-conference workshops and the face-to-face conference, you will achieve the "CDG Expert Diploma". Stay tuned!

- **World CDG Advocacy and Leadership Academy**, 20-21 July 2023: If you are beginning to think about advocacy and do not have any experience so far, please do not worry. "World CDG Advocacy and Leadership Academy" is delivered in English and includes assistance to the virtual "World Think Metabolic, Think CDG Academy" and in-person sessions before and during the 6th World Conference on CDG.
  "World CDG Advocacy and Leadership Academy" is tailored for leaders of CDG patient groups and their advocates and helps our CDG Organizations and representatives to be successful advocates.
  We also offer courses to help with everyday life with CDG. You can meet CDG advocates and those making a difference for people with CDG every day!
  Participants will acquire knowledge and skills to participate in research and drug development activities with all stakeholders in a collaborative and global mindset.
- **6th World Conference on CDG**, 21-23 July 2023 takes place from 21 July to 23 July 2023. We are investigating the possibility of simultaneous translation during the in-person conference in Caparica, Portugal.

Learn more here
A Webinar aimed at sharing novelties about the 6th World Conference on CDG (WCCDG) 2023, including the "World Think Metabolic, Think CDG Academy" and "World CDG Advocacy and Leadership Academy" will be held 20 January 2023 from 16-17 PM (GMT), online and in English.
Sign up at WorldCDG.org

Evidence Based Advocacy

Why "Evidence-Based Patient Advocacy"?

People living with a certain condition and their advocates may best communicate their needs. However, individual perspectives are not necessarily the most effective to influence researchers, healthcare practitioners, or regulatory decision-makers. In order to encourage healthcare to focus on the genuine needs of patients and their subpopulations, patient advocates should create rigorous evidence and share it.

What is "Evidence-Based Patient Advocacy"?

"Evidence-Based Patient Advocacy" involves advocating in a targeted, scientific, well-educated, and professional manner to quantify the impact of advocacy activity. It is built on three main elements:

- targeted advocacy towards each respective stakeholder,
- use of robust data about patients' needs and preferences,
- and use of the correct packaging of messages to express the needs to the respective target group.

Which challenges do patient organisations face when they wish to generate data for advocacy?

There are several challenges:

- the capacity to undertake an evidence-based project in a methodological manner
- the knowledge of how to utilize data in advocacy
- the knowledge, capability, and resources to disseminate the data
- a strategy to ensure data generation is executed only when invited/encouraged by industry

What kind of meaningful data can be generated by patient groups?

- Adherence to therapies
- Inequalities in real-world access to diagnostics or therapies
- Current care patterns
- Quality of Life, burden of disease, daily lived experience in real world
- Impact of illness on society
- Disease-related outcomes
- Patient preferences on benefit/risk

Learn more about "Evidence-Based Patient Advocacy" here

An Evidence-Based Patient Advocacy best practice in CDG transferable across rare diseases: The CDG Journey Mapping

The journey through life with a rare disease

When a person seeks medical care for symptoms, the diagnostic journey begins and ends with a correct diagnosis or a regular checkup to receive treatment for an illness or injury. Nowadays, thanks to advances in modern medicine, we could expect that a person can quickly receive a diagnosis and treatment. Those families living with rare conditions often spend years or decades seeking a diagnosis. **Rare conditions take an average of 6 to 7 years to diagnose**. People living with rare diseases may see eight or more doctors and undergo extensive testing before receiving a final diagnosis. Families and providers often get frustrated during long diagnostic journeys. Those living with a rare condition and caregivers may feel alone. Additionally, there are burdens of cost, intensive utilization of resources, and many other challenges. Rare Disease International made available the following explanatory video about the journey through life with a rare disease:

What is the CDG Journey Mapping?

The study CDG Journey Mapping is the first of its kind, providing the most comprehensive understanding of the entire sequence of events that a patient experiences within a given healthcare system or across providers, from initial signs and symptoms to receiving a definitive diagnosis or treatment for an illness or injury. Learn more here.

Why is it important to understand the CDG patient Journey

Our research team has identified a major gap in current CDG knowledge. The CDG journey is still unknown. We have all heard about family's experiences and views when questing for a diagnosis and fighting for adequate care for their beloved ones. But there is no systematic study in CDG that captures the various aspects of the CDG family journey in a robust manner.
What are their concerns, priorities, requirements, needs, and expectations when living with CDG? Which symptoms have the most significant impact on their quality of life? What are the current CDG management and care strategies? How usually a family deals with the diagnosis? How long does it take to get a definitive diagnosis? All of these questions, alongside a lot of others, are addressed by developing and refining a structured CDG Journey Mapping.

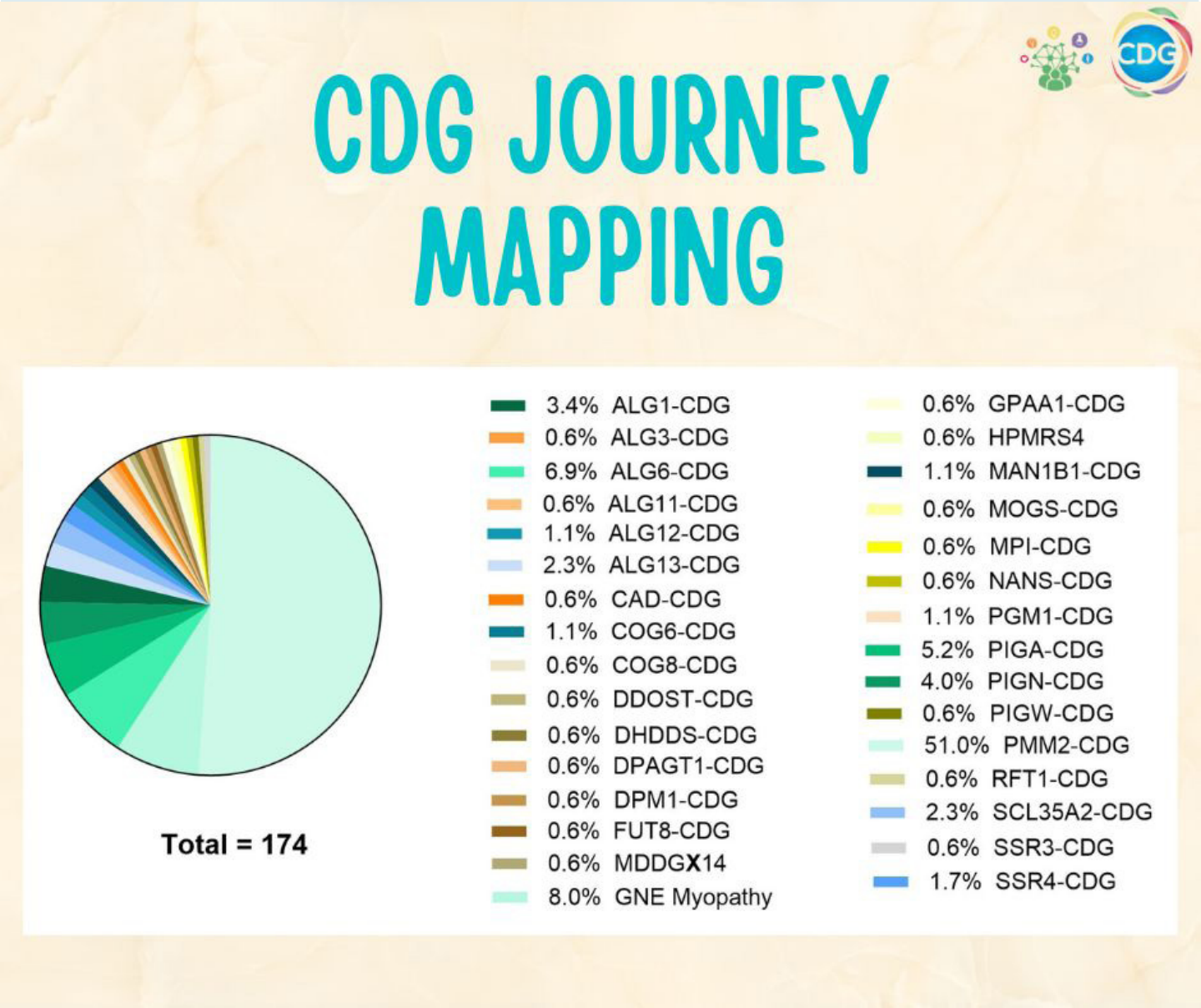
Results from the CDG Journey Mapping questionnaire

The CDG community gave us this amazing gift by responding to our questionnaires. This just shows how wonderful research partners CDG families and professionals are!
Do you want to have a glimpse of the results? Here it goes the first results from the CDG Journey Mapping questionnaire! Learn everything here.

The TOP 4 answers to the following question "Which CDG type affects you/your child/relative?" are:

- 89 PMM2-CDG
- 14 GNE-Myopathy
- 12 ALG6-CDG
- 10 PIGA-CDG

Look at the image below.



Discover more about the CDG Journey Mapping on our resources.

In a rare disease community your voice values gold, MAKE YOUR VOICE BE HEARD!

News from CDG research networks

The CDG and Allies Community-centric International Research Network

What is CDG & Allies?

With the help of a broad network of scientists, physicians, families and patient advocacy groups, we have established a patient-led national and internationally unrivaled infrastructure for research, awareness and education for CDG. The research on Glycosylation disorders is primarily dedicated to Congenital Disorders of Glycosylation (CDG). The advances and innovations achieved for CDG through CDG & Allies - PPAIN will impact on a large number of patients. Namely, the overall human diseases characterized by abnormal protein glycosylation such as cancer, inflammation, Alzheimer's disease and diabetes.

CDG & Allies – PPAIN
CDG & Allies - Professionals and Patient Associations International Network

What are the specific objectives?

- To translate the expertise of our collaborators for the benefit of CDG.
- To boost accurate and timely diagnosis.
- To enable educational and training initiatives.
- To potentiate active exchange of experiences and best practices within key stakeholders.
- To develop partnerships.
- To empower families as partners through accurate information and educational material.
- To develop sustainability by identifying options for funding.

Our research model:

Fomenting the research on rare diseases is essential to pave the way for therapeutic approaches and development, since for most of the patients there is still no treatment. In addition, experience says that any knowledge on the mechanism underlying rare diseases and its pathological manifestations will be beneficial, not only for those patients but also other patients with related pathological symptoms. Our patient-initiated research model:

The Frontiers in Congenital Disorders of Glycosylation Consortium (FCDGC's)

About the Consortium

The FCDGC is part of the Rare Diseases Clinical Research Network (RDCRN), which is funded by the National Institutes of Health (NIH) and led by the National Center for Advancing Translational Sciences (NCATS) through its Division of Rare Diseases Research Innovation (DRDRI).

Video: Pf Eva Morava-Kozicz, MD, PhD of the Mayo Clinic in Rochester, MN profiles an overview of congenital disorders of glycosylation (CDG).

Frontiers in Congenital Disorders of Glycosylation Consortium (FCDGC's) vision is to conduct research to support the best integrated clinical care for congenital disorders of glycosylation (CDG) in the nation.

A strong patient association, committed clinicians and a devoted group of scientists has formed a virtual consortium over the last decade and have closely collaborated to improve patient outcomes. Partners in our consortium have collaborated for many years—sharing knowledge, individualizing therapy, organizing patient conferences, and supporting physicians caring for individuals with CDG patients. We have improved patient care in this rare disease. On this foundation we have set out to establish a nationwide network of regional centers to relieve decades of unresolved questions, address knowledge gaps, develop treatment and meet currently unmet patient needs.

FCDGC

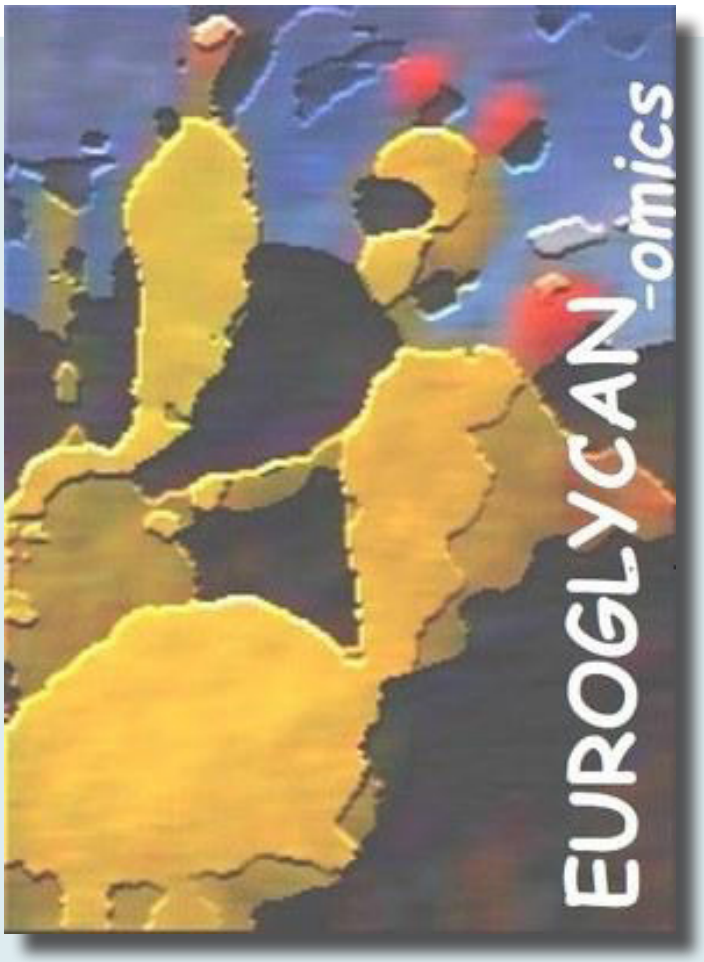
RARE DISEASES
CLINICAL RESEARCH NETWORK

Consortium History

Congenital disorders of glycosylation (CDG) consist of more than 170 different inborn errors of metabolism at an estimated overall incidence of greater than 1 in 100,000. While these disorders were first genetically defined in the 1990s, there is no data available on their natural history, no comprehensive patient registry, no reliable screening tests for many types, and large gaps in clinical trial readiness. In response, a nationwide network of 13 sites total (including 9 clinical sites) was established to: define the natural history; validate patient-reported outcomes and share CDG knowledge; develop and validate new biochemical diagnostic techniques and therapeutic biomarkers to increase clinical trial readiness; and evaluate whether dietary treatments restore appropriate glycosylation to improve clinical symptoms and quality of life.
As with all RDCRN consortia, patients are key partners in research.

The consortium leverages cross-disciplinary, team-based clinical science to address decades of unresolved questions; increase clinical trial readiness; advance and share knowledge, awareness and education on CDG; and, most importantly, develop treatments and meet unmet patient needs.

The Euroglycan-omics



Glycosylation is a chemical modification of proteins in our cells. It affects their stability and function. Thousands of proteins are 'glyco-proteins'. As a result, genetic defects in glycosylation pathways lead to mostly severe diseases, called 'Congenital Disorders of Glycosylation' (CDG). With more than 160 different types, CDG has become an impressive group of metabolic diseases.

On the one hand, we have patients with clear glycosylation anomalies, whose genetic defects remain elusive. We plan to use genomic tools to identify new genes and new disease mechanisms. On the other hand, we believe that many other patients with symptoms that fit with CDG (like intellectual disabilities, multiple congenital anomalies, bleeding disorders etc.), may have a glycosylation disorder that presently escapes diagnosis because the diagnostic tools are insufficient. We will develop new biomarkers based on glycomics, glycopeptidomics and metabolomics. In addition, fundamental research into the biochemistry of the sugars may lead to the identification of completely novel types of diseases.
The ultimate goal is to treat or cure CDG patients. Since the cellular compartments where glycosylation occurs cannot easily be reached with therapeutic compounds, a causative treatment is difficult. Therefore, a better understanding of the pathophysiology of the different types of CDG is crucial for the development of novel therapeutic interventions. To this end we will systematically study already available cellular and mouse models using state-of-the-art omics approaches.
EUROGLYCAN-omics brings together a multidisciplinary group of clinical and basic researchers who have proven to be able to collaborate. We want to achieve the ambitious goals set here, for the sake of the patients and their families.
More information: here.

Glycomine GLM101 Substrate Replacement Therapy in Adults Subjects with PMM2-CDG (NCT05549219)

The GLM101 clinical trial for adult PMM2-CDG patients is now recruiting in Spain, Australia, and the United States.
This study is designed to assess the safety, tolerability, pharmacokinetics and biochemical changes of adult subjects with PMM2-CDG after 3 months of treatment with GLM101.
The study is seeking 6 patients, ages 18-65 years, for enrollment. Study centers are located in the United States (Central Research of Florida, Tampa, FL; Mayo Clinic, Rochester, MN; University of Minnesota, MN), Australia (Royal Adelaide Hospital, Adelaide, South Australia; Royal Melbourne Hospital; Victoria, Australia) and Spain (San Joan de Deu, Barcelona).

Glycomine and the study centers will manage travel reimbursement and coordination of study visit dates.
To know more about Glycomine's method to increase the uptake of mannose -1-phosphate by the patient's body, watch the video below "Designing a potential treatment for PMM2-CDG".

Learn more here
To learn more about this trial, or to schedule your appointment, please contact one of the study centers listed on www.clinicaltrials.gov or e-mail info@glycomine.com.

Clinical Trials

According to clinicaltrials.gov under the U.S. National Institutes of Health, the EU Clinical Trials Register (EU-CTR), and the International Clinical Trials Registry Platform (ICTRP) under the World Health Organization (WHO) **the following trials should be recruiting at this moment:**

- Dietary Monosaccharide Supplementation in Patients With Congenital Disorders of Glycosylation
- Galactose Supplementation for the Treatment of MOGHE
- Oral Epalrestat Therapy in Pediatric Subjects With PMM2-CDG
- AVTX-801 D-galactose Supplementation in SLC35A2-CDG
- Evaluate Optimal Dosing and Long-term Safety of D-galactose in PGM1-CDG
- Clinical and Basic Investigations Into Known and Suspected Congenital Disorders of Glycosylation
- Clinical and Basic Investigations Into Congenital Disorders of Glycosylation
- Trial of lactose supplementation in CDG
- Investigation of the underlying pathomechanisms found in defects of the neurotransmitter, Pterine -, phenyl alanine, and 5-Methyltetrahydrofolate metabolism in induced pluripotent stem cells (iPSC) and derivatives
- "Incidence and Consequences of Disorders of Glycosylation in Patients With Conotruncal and Septal Heart Defects"

The following trials **are not yet recruiting at this moment:**

- Evaluate the Effect of Oral GlcNAc Supplementation in Patients With NGLY1 Deficiency
- 12-Week Study to Assess the PD, Safety, Tolerability, and PK of GLM101 in Adults With PMM2-CDG (as shared in the previous section dedicated to News from industry, some sites are now recruiting and this information should be updated soon at www.clinicaltrials.gov)

The following trials **are active, not yet recruiting:**

- Acetazolamide Efficacy in Ataxia in PMM2-CDG

The following **are enrolling by invitation**:

- Large-Scale Metabolomic Profiling for the Diagnosis of Inborn Errors of Metabolism

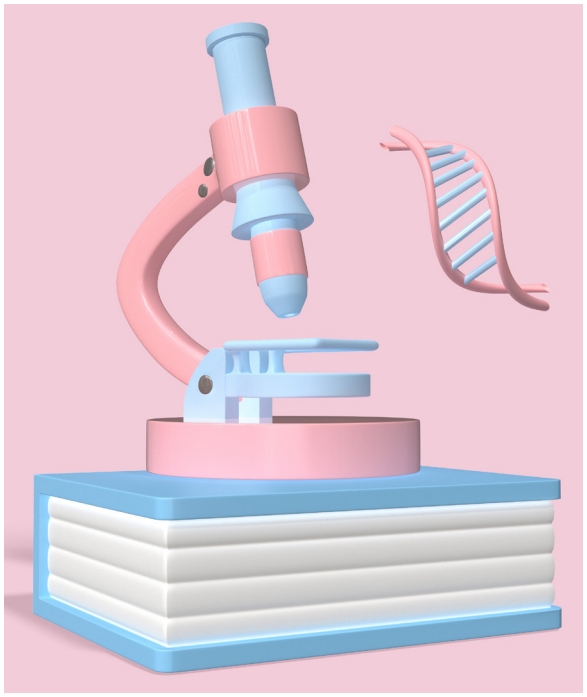
To search for clinical trials go to www.clinicaltrials.gov, or International Clinical Trials Registry Platform or EU Clinical Trials Register.

To better understand clinical trials, read the sections available at WorldCDG.Org

- I want to learn about Clinical Trials
- I want to participate in Clinical Trials
- I am a Clinical Trial participant
- Considering a clinical trial
- Questions to ask your Clinical team
- Frequently Asked Questions

Here are some of the recently published CDG related scientific papers available thanks to Pubmed:

- Paprocka J. Neurological Consequences of Congenital Disorders of Glycosylation. Adv Neurobiol. 2023;29:219-253. doi: 10.1007/978-3-031-12390-0_8. PMID: 36255677.
- Wenzel DM, Olivier-Van Stichelen S. The O-GlcNAc cycling in neurodevelopment and associated diseases. Biochem Soc Trans. 2022 Nov 16:BST20220539. doi: 10.1042/BST20220539. Epub ahead of print. PMID: 36383066.
- Ng BG, Sosicka P, Xia Z, Freeze HH. GLUT1 is a Highly Efficient L-Fucose Transporter. J Biol Chem. 2022 Nov 21:102738. doi: 10.1016/j.jbc.2022.102738. Epub ahead of print. PMID: 36423686.
- Mitchell CW, Czajewski I, van Aalten DMF. Bioinformatic prediction of putative conveyers of O-GlcNAc transferase intellectual disability. J Biol Chem. 2022 Sep;298(9):102276. doi: 10.1016/j.jbc.2022.102276. Epub 2022 Jul 19. PMID: 35863433; PMCID: PMC9428853.
- Valko A, Gallo GL, Weisz AD, Parodi AJ, D'Alessio C. Analysis of Lipid-linked Oligosaccharides Synthesized in vivo in Schizosaccharomyces pombe. Bio Protoc. 2022 Sep 20;12(18):e4508. doi: 10.21769/BioProtoc.4508. PMID: 36311347; PMCID: PMC9550348.
- Masunaga Y, Nishimura G, Takahashi K, Hishiyama T, Imamura M, Kashimada K, Kadoya M, Wada Y, Okamoto N, Oba D, Ohashi H, Ikeno M, Sakamoto Y, Fukami M, Saitsu H, Ogata T. Clinical and molecular findings in three Japanese patients with N-acetylneuraminic acid synthetase-congenital disorder of glycosylation (NANS-CDG). Sci Rep. 2022 Oct 12;12(1):17079. doi: 10.1038/s41598-022-21751-x. PMID: 36224347; PMCID: PMC9556533.
- Caino S, Cubilla MA, Alba R, Obregón MG, Fano V, Gómez A, Zecchini L, Lapunzina P, Aza-Carmona M, Heath KE, Asteggiano CG. Clinical and Genetic Analysis of Multiple Osteochondromas in A Cohort of Argentine Patients. Genes (Basel). 2022 Nov 7;13(11):2063. doi: 10.3390/genes13112063. PMID: 36360300; PMCID: PMC9690389.



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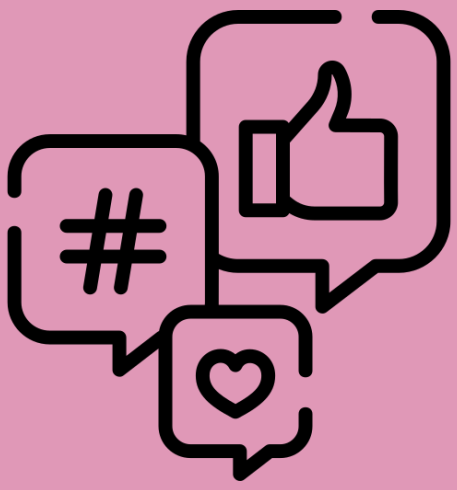

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©World CDG Magazine, created and written by Vanessa Ferreira, Senior people-centric researcher and co-founder of the international research network CDG and Allies, Founder of the Portuguese Association for CDG (APCDG), Global Patient Advocate, and sibling to Princess Liliana.

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