

5th World Conference on CDG for families and professionals

ONLINE EVENT, 13-16 MAY 2021

#WCCDG2021

Organised by the Portuguese Association for CDG (APCDG) and CDG & Allies Professionals and Patient Associations International Research Network (CDG & Allies PPAIN), and in collaboration with our Global CDG community (families, patient groups and professionals).

Copyright © 2021 APCDG.

*****THE FINAL PROGRAM*****

5th World Conference on CDG for families and professionals

The multidisciplinary scientific forum to advance and disseminate knowledge in the field WITH and FOR CDG families and professionals

***** PROGRAM*****

Follow our News about the congress at <https://worldcdg.org/world-conference-cdg>

Conference Theme: “#StandUnited4CDG People Centric approach that drives CDG therapeutic development: Impossible, Is Possible!”

When: Thursday 13 to Sunday 16 May 2021

How: This edition, will exceptionally be virtual due to the COVID-19 pandemic.

Key Dates

Due to COVID-19 uncertainties, the congress needs early anticipation and preparation, thus please take the following congress dates into consideration. And secure your online attendance at the earliest!

Dates to remember	When
Registration	1st March -1st April 2021
Extension for registration	2nd-9th April 2021
ePoster Abstract Submission	1st March -1st April 2021
Extension ePoster Abstract Submission	1st March -1st April 2021
Notification of poster abstracts accepted for display at congress website and oral presentation	2nd - 16th April 2021
ePoster PDF upload at https://worldcdg.org/world-conference-cdg/upload-your-e-poster-pdf	26th April 2021
Oral presentation Video recorded ePoster to be sent to organisers (it is strongly recommended you follow guidance sent to you by email)	30th April 2021

World Conference on CDG	13-16 May 2021
World Awareness CDG Day 2021	16 May 2021

Major Novelty: The majority of the conference sessions enter under the scope of the **“Think Metabolic, Think CDG” Academy** for families, academia, medical doctors and industry. The **“Think Metabolic, Think CDG” Academy** aims to provide CDG stakeholders the knowledge and skills needed to become experts in CDG medicines research and development. Thus, by securing your attendance and actively participating during the conference, you will be able to achieve the “CDG Expert Level 2”. Further details, **SOON! Stay tuned!**

Day 1: Thursday 13th May 2021

Simultaneous interpretation today is offered in the following languages:

- English > Portuguese
- English> Italian

Thursday 13th May 2021 from 16:30 – 17:00 PM Lisbon (Portugal).

Want to know what time corresponds in your country? Use World Time Buddy (WTB) at

<https://www.worldtimebuddy.com/>

Virtual gathering to Secure you connect with some anticipation.

Theme 1: Actions to boost CDG research and drug development.

**Thursday 13th
May 2021**

**17:00 - 17:05
PM Lisbon
(Portugal)**

Welcome to the 5th World Conference CDG by organisers.

Carlota Pascoal, Rita Francisco, Paula Videira and Vanessa Ferreira (Portugal).

Thursday 13th May 2021 17:05 - 19:00 PM Lisbon (Portugal) (10 Minutes biobreak is included)	Introduction and session presentation by Moderator(s) (8 minutes): Inês Alves (Portugal), Rita Francisco (Portugal) and Vanessa Ferreira (Portugal). Community CDG Think Tank 1: The challenges and solutions faced throughout CDG research and drug development: Results from an international research study made possible by CDG families and professionals. Talk Title and corresponding Speaker(s) 1 (15 minutes): The challenges and solutions faced throughout CDG research and drug development: Results from an electronic questionnaire made possible by CDG families and professionals, by Maria Monticelli (Italy). Round table discussion with Panelists facilitated by Moderator(s) (15 minutes): Inês Alves (Portugal), Rita Francisco (Portugal) and Vanessa Ferreira (Portugal). Moderators supported by: Bruna Henriques, Madalena Parrado and Susana Alves from Sci and Volunteer Program FCT NOVA 2021. Talk Title and corresponding Speaker(s) 2 (15 minutes): Identifying the challenges and solutions of therapy in Congenital Disorders of Glycosylation (CDG): A multi-stakeholder, mixed-research approach by Rita Francisco (Portugal) and Vanessa Ferreira (Portugal). Round table discussion with Panelists facilitated by Moderator(s) (35 minutes): Inês Alves (Portugal), Rita Francisco (Portugal) and Vanessa Ferreira (Portugal). Moderators supported by: Bruna Henriques, Madalena Parrado and Susana Alves from Sci and Volunteer Program FCT NOVA 2021. Panelists: Poster presenters are also invited to share views during the round table.
---	--

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Bobbie List (USA)
- Family and/or Patient Group Perspective: Julie Zeh (USA)
- Family and/or Patient Group Perspective: Yolandi Botha (South Africa)
- Family and/or Patient Group Perspective: Wendy Kirts (USA)
- Family and/or Patient Group Perspective: Allison S. Hartnett (USA)
- Researcher Perspective: Paula Videira (Portugal)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Researcher Perspective: Javier Corral (Spain)
- Researcher Perspective: Cecilia D'Alessio (Argentina)
- Clinician Perspective: Teona Shatirishvili (Georgia)
- Clinician Perspective: Tamas Kozicz (USA)
- Pharmaceutical industry Perspective: John Boland (Cerecor, USA)

BIOBREAK 10 minutes

Lightning to Posters session:

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes per poster presentation):

- Spontaneous improvement of carbohydrate-deficient transferrin in PMM2-CDG by Christin Johnsen (Mayo Clinic, USA)
- SLC35A2-CDG: diagnosis and new therapeutic approaches by Bibiana Mello de Oliveira (HCPA, UFRGS, HCSA, Mendelics, Brasil)

- Why and how do we need to measure sugar metabolism in CDG? by Raisa Veizaj (Radboudumc, The Netherlands)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~15-20 minutes):

Inês Alves (Portugal), Rita Francisco (Portugal) and Vanessa Ferreira (Portugal).

Moderators supported by: Bruna Henriques, Madalena Parrado and Susana Alves from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Special Rare Disease Expert:

Inês Alves (Portugal) (this expert when adequate, will share learnings and best practices from another Rare Disease area named Achondroplasia that can be transferred for CDG families and professionals across countries).

Session Brief description:

Community CDG Think Tank 1: The challenges and solutions faced throughout CDG research and drug development: Results from an international research study made possible by CDG families and professionals.

Back to the 4th World CDG Congress in 2019, an international electronic survey research study was held. It was aimed at identifying challenges and solutions throughout CDG research and drug development from families and professionals'

	views. This session will show the Why's and How's of efficiently integrating the voice and experience of people living with CDG and professionals, throughout research and drug development. The results will be shared for the first time., and major points will be discussed including: • Overview of CDG needs and solutions to stimulate future CDG research and drug development • Top ten actions to boost CDG research and drug development • Identify and assess best practices that can be transferable for CDG across countries • Identify united the 3 short- & 3 long-term priority solutions to tackle the challenges identified • Ways to measure implementation of priority solutions
Thursday 13th May 2021 from 19:00 – 19:15 PM, Lisbon, Portugal BREAK	

Day 1: Thursday 13th May 2021

Simultaneous translation today is offered in the following languages:

- English > Portuguese
- English> Italian

Theme 1: Actions to boost CDG research and drug development.

Thursday 13th May 2021	Introduction and session presentation by Moderator(s) (8 minutes): Rita Francisco (Portugal) and Vanessa Ferreira (Portugal)
19:15 – 21:00 PM, Lisbon, Portugal	Community CDG Think Tank 2: Families experiences when managing CDG symptoms: care, management, rehabilitation therapies, diets and impact in daily life.
(10 Minutes break is included)	Talk Title and corresponding Speaker(s) (15 minutes): Prioritizing Symptoms Impacting Quality of Life for Congenital Disorders of Glycosylation (Part 1 of the talk) by Cátia Neves (Portugal).
	Round table discussion with Panelists facilitated by Moderator(s) (25 minutes): Rita Francisco (Portugal) and Vanessa Ferreira (Portugal).
	Moderators supported by: Bruna Henriques, Madalena Parrado and Susana Alves from Sci and Volunteer Program FCT NOVA 2021.

BIOBREAK 10 Minutes

Talk Title and corresponding Speaker(s) (15 minutes):

Prioritizing Symptoms Impacting Quality of Life for Congenital Disorders of Glycosylation (Part 2 of the talk) by Cátia Neves (Portugal).

Round table discussion with Panelists facilitated by Moderator(s) (25 minutes):

Rita Francisco (Portugal) and Vanessa Ferreira (Portugal).

Moderators supported by: Bruna Henriques, Madalena Parrado and Susana Alves from Sci and Volunteer Program FCT NOVA 2021.

Panellists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Kayla Card (Australia)
- Family and/or Patient Group Perspective: Mariana Esquinca (México)
- Family and/or Patient Group Perspective: Megan Bonavia (Australia)
- Family and/or Patient Group Perspective: Brea McClain (USA)
- Family and/or Patient Group Perspective: Wanda Kadziolka (Canada)
- Researcher Perspective: María Eugenia de la Morena-Barrio (Spain)
- Researcher Perspective: Oriol Gallego (Spain)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Researcher Perspective: Kristin Kantautas (Canada)
- Clinician Perspective: Peymaneh Sarkhail (Irán)
- Clinician Perspective: Miriam Janssen (The Netherlands)

- Clinician Perspective: Mercedes Serrano (Spain)
- Pharmaceutical industry Perspective: Jahannaz Dastgir (Applied Therapeutics, USA)
- Pharmaceutical industry Perspective: Joff Masukawa (Cerecor, USA)

Special Rare Disease Expert:

Femida Gwadry-Sridhar (this expert from Pulse infoframe will share learnings and best practices from another Rare Disease area that can be transferred for CDG families and professionals across countries).

Lightning to Posters session:

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes per poster presentation):

- Generation of cellular models to study congenital disorders of glycosylation by Rachel Mijdam (Radboudumc, The Netherlands)
- Cellular and animal models to study immune cell functions in CDG, by Alessandra Cambi (Department of Cell Biology, Radboud university medical center, The Netherlands)
- Zebrafish as a model for CDG diseases, by Nerea Gandoy Fieiras (Department of Zoology, Universidade de Santiago de Compostela, Spain)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~15 minutes):

Rita Francisco (Portugal) and Vanessa Ferreira (Portugal).

Moderators supported by: Bruna Henriques, Madalena Parrado and Susana Alves from Sci and Volunteer Program FCT NOVA 2021.

- Attendees: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.

- Panelists & poster presenters: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

Community CDG Think Tank 2: Families experiences when managing CDG symptoms: care, management, rehabilitation therapies, diets and impact in daily life.

CDGs are often characterised by complex pathophysiology, a wide range of symptoms and outcomes, and a poorly understood experience from people living with CDG and their family members views. Signs and symptoms can cause functional limitations that may worsen over the disease trajectory and may impact people living with CDG and family members quality of life. Currently, any study has systematically examined the impact of the core set of CDG signs/ and symptoms. Also, there is not collective evidence of current management, rehabilitation therapies and diets. This knowledge is the first step to ensure that clinical trials and therapeutic interventions meet the expectations of people living with CDG and their family members, needs and, hopefully, lead to improved outcomes for people living with CDG.

Early 2021, an international research study led by CDG & Allies PPAIN in collaboration with worldwide CDG families, patient groups and professionals was conducted and you will hear first-hand collective results from this international research. This session will walk you throughout the:

- Which CDG signs and symptoms present major frequency and severity in family's daily lives?
- List of CDG signs and symptoms that families consider priority to treat
- Managing CDG signs and symptoms (medication, diets and rehabilitation therapies)
- Main causes of CDG hospitalizations and impact
- Outline of CDG care specialists and reference centres across countries
- CDG childhood to adulthood transition across countries
- The impact CDG signs and symptoms in the daily life of people living with CDG and caregiver's activities/tasks

Day 2: Friday 14th May 2021

Simultaneous translation today is offered in the following languages:

- English > Portuguese
- English> Italian

Friday 14th May 2021 from 14:00 -14:20 PM, Lisbon, Portugal.

Want to know what time corresponds in your country? Use World Time Buddy (WTB) at

<https://www.worldtimebuddy.com/>

Virtual gathering to Secure you connect with some anticipation.

**14:20-14:30
PM, Lisbon,
Portugal**

Welcome to the 5th World Conference CDG by organisers.
Carlota Pascoal, Rita Francisco, Paula Videira and Vanessa Ferreira (Portugal).

Theme 2: CDG Classification and Diagnosis: present, needs and solutions.

**Day 2: Friday
14th May 2021**

Introduction and session presentation by Moderator(s) (8 minutes):
Carlos Ferreira (USA), Rita Francisco (Portugal) and Carlota Pascoal (Portugal).

14:30 – 16:00 PM, Lisbon, Portugal	Keynote session 1: CDG a growing family – New CDG types and update on CDG classification. Talk Title and corresponding Speaker(s) 1 (15 minutes): CDG: A tale with many branches and numerous twists, by Rita Francisco (Portuguese Association for CDG and CDG & Allies PPAIN, Portugal) by Rita Francisco (Portugal). Round table discussion with Panelists facilitated by Moderator(s) (20 minutes): Carlos Ferreira (USA), Rita Francisco (Portugal) and Carlota Pascoal (Portugal). Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021. Talk Title and corresponding Speaker(s) 2 (13 minutes): Epidemiology of Congenital Disorders of Glycosylation (CDG), by Ana Piedade (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal). Round table discussion with Panelists facilitated by Moderator(s) (15 minutes): Carlos Ferreira (USA), Rita Francisco (Portugal) and Carlota Pascoal (Portugal). Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021. Panelists: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. • Family and/or Patient Group Perspective: Mandy Pinheiro (South Africa) • Family and/or Patient Group Perspective: Sanja Juric (Germany) • Family and/or Patient Group Perspective: Julia Boonak (CDG UK)
--	--

- Family and/or Patient Group Perspective: Kara Berasi (USA)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Researcher Perspective: Rita Francisco (Portugal)
- Clinician Perspective: Carlos Ferreira (USA)
- Clinician perspective: Jaak Jaeken (Belgium)
- Clinical Perspective: Irakli Rtskhiladze (Georgia)

Lightning to Posters session

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes per poster presentation):

- EDEM3-CDG, a new congenital disorder of glycosylation comprising non-specific intellectual disability, by Andrew Edmondson (Children's Hospital of Philadelphia, USA)
- Genotypic and phenotypic spectrum in Chinese patients with congenital disorders of glycosylation, by Kuerbanjiang Abuduxikuer (Department of Hepatology, Children's Hospital of Fudan University, National Children's Medical Center, China)
- Insight into patient reported experiences diagnosed with Congenital Disorders of Glycosylation (CDG), by Andrea Miller (CDG USA)
- The prevalence of congenital disorders of glycosylation in childhood epilepsy, by Saadet Mercimek-Andrews (Department of Medical Genetics, University of Alberta, Canada)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~10 minutes):

Carlos Ferreira (USA), Rita Francisco (Portugal) and Carlota Pascoal (Portugal).

Moderators supported by: Marta Silva, Madalena Abade, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

Keynote session 1: CDG a growing family – New CDG types and update on CDG classification.

Since their first clinical description in 1980, approximately 170 types of CDG have been identified, and that number keeps rising. CDG clinical spectrum is extremely broad, covers nearly all known phenotypes, and comprises new phenotypes. This session will cover:

- Why is CDG classification and nomenclature important?
- How is CDG classification done?
- CDG types and update on CDG classification with main associated organ involvement and symptomatology
- What are the 3 top challenges and solutions associated with CDG classification and nomenclature from CDG stakeholders' views?

16:00 – 16:15 PM, Lisbon, Portugal

BREAK

Day 2: Friday 14th May 2021 16:15 – 17:30 PM, Lisbon, Portugal	Introduction and session presentation by Moderator(s) (8 minutes): Vanessa Ferreira (Portugal), Pedro Granjo (Portugal), Rita Francisco (Portugal) and Carlota Pascoal (Portugal). Panel of discussion 1: The quest from first manifestations to final CDG diagnosis: needs, hopes and best practices from families and professionals’ perspectives. Talk Title and corresponding Speaker(s) (15 minutes): The quest from first manifestations to final CDG diagnosis: needs, hopes and best practices from families and professionals’ perspectives, by Vanessa Ferreira (Portugal) and Pedro Granjo (Portugal) from Sci and Volunteer Program FCT NOVA 2021. Round table discussion with Panelists facilitated by Moderator (s) (30 minutes): Vanessa Ferreira (Portugal), Pedro Granjo (Portugal), Rita Francisco (Portugal) and Carlota Pascoal (Portugal). Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. • Family and/or Patient Group Perspective: Louise Ward (UK) • Family and/or Patient Group Perspective: Heather Conneran (USA) • Family and/or Patient Group Perspective: Megan Winkels DeHaven (USA) • Family and/or Patient Group Perspective: Sophie Clayton (UK) • Researcher Perspective: Hudson Freeze (USA)
---	---

- Researcher Perspective: Kimiyo Raymond (USA)
- Researcher Perspective: Maria Isabel Quiroga (Spain)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Researcher Perspective: Dirk Lefeber (The Netherlands)
- Clinician Perspective: Tinatin Tkemaladze (Georgia)
- Clinician Perspective: Agata Fiumara (Italy)

Special Rare Disease Expert:

Rebecca Stewart, Rare Revolution Magazine (UK) (this expert will share learnings and best practices from another Rare Disease area that can be transferred for CDG families and professionals across countries).

Lightning to Posters session

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes per poster presentation):

- FUT8-CDG explained: An informative, and empowering community-friendly resource, by Heather Conneran (CDG CARE USA) & supported by Carlota Pascoal (Portuguese Association for CDG and CDG & Allies PPAIN, Portugal)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~20 minutes):

Vanessa Ferreira (Portugal), Pedro Granjo (Portugal), Rita Francisco (Portugal) and Carlota Pascoal (Portugal).

Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

The difficulty in obtaining the correct diagnosis is still the first dramatic hurdle for many CDG families and may take years to overcome. CDG diagnosis is a challenge, not only because of the large number of CDGs types but also because of the huge clinical heterogeneity even within a number of CDG. So far, no systematic study about information needs at the time of the announcement of CDG diagnosis has been done. An international research study named **CDG Journey Mapping**, is **currently** led by CDG & Allies PPAIN in collaboration with worldwide CDG patient groups, families and professionals. It aims to collect the quest from first manifestations to final CDG diagnosis as well as immediate needs, hopes and best practices transferable for CDG across countries from families and professionals' perspectives.

Several results will be covered in this session, such as:

- What is the quest for a correct CDG final diagnosis?
- What are the current challenges related to CDG biochemical and genetic testing?
- Which new approaches and existing best practices can improve CDG diagnosis?
- What is the role of CDG patient groups in providing information about CDG at the time of diagnosis?
- What are the needs and solutions from families and professionals' views related to CDG information at the time of diagnosis?
- List of information and support best practices at the time of diagnosis and transferable across countries.

Day 2: Friday 14th May 2021 from 17:30 – 17:45 PM, Lisbon, Portugal

BREAK

Day 2: Friday 14th May 2021 17:45 -19:00 PM, Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Maria Grázia Rossi (Portugal) and Rita Francisco (Portugal) Panel of discussion 2: CDG families—physician communication and short- and long-term support after diagnosis: families and professionals views and experiences. Talk Title and corresponding Speaker(s) 1 (12 minutes): Be a CDG Health Literacy Hero using WorldCDG.Org Now! The CDG at the forefront of Plain Language resources and framework, by Salvador Magrinho (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal). Talk Title and corresponding Speaker(s) 2 (4 minutes): Plain Language of Publications (PLPs): helping disseminate published scientific articles to people living with CDG and empowering the community, by Marta Abreu (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal). Round table discussion with Panelists facilitated by Moderator (s) (15 minutes): Maria Grázia Rossi (Portugal) and Rita Francisco (Portugal). Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.
--	---

- Researcher Perspective: Dulce Quelhas (Portugal)
- Family and/or Patient Group Perspective: Ananias Gonzalez (Spain)
- Family and/or Patient Group Perspective: Amy Dann (Australia)
- Family and/or Patient Group Perspective: Femke van der Maat – de Deugd (The Netherlands)
- Family and/or Patient Group Perspective: Marta Kadziolka (Canada)
- Researcher Perspective: Marta Falcão (Portugal)
- Researcher Perspective: Bobby G Ng (USA)

Talk Title and corresponding Speaker(s) 3 (15 minutes):

The road to successful people-centric research in rare diseases: The web-based case study of the Immunology and Congenital Disorders of Glycosylation questionnaire (ImmunoCDGQ) by Rita Francisco.

Talk Title and corresponding Speaker(s) 4 (4 minutes):

Empowering CDG families and professionals with an arsenal of educational resources, by Marta Falcao (NOVA School of Science and CDG & Allies PPAIN, Portugal).

Round table discussion with Panelists facilitated by Moderator (s) (10 minutes):

Maria Grázia Rossi (Portugal) and Rita Francisco (Portugal).

Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021.

Panelists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Ananias Gonzalez (Spain)

- Family and/or Patient Group Perspective: Amy Dann (Australia)
- Family and/or Patient Group Perspective: Femke van der Maat – de Deugd (The Netherlands)
- Family and/or Patient Group Perspective: Marta Kadziolka (Canada)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Researcher Perspective: Marta Falcão (Portugal)
- Researcher Perspective: Bobby G Ng (USA)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~10 minutes):

Maria Grázia Rossi (Portugal) and Rita Francisco (Portugal).

Moderators supported by: Marta Silva, Madalena Abade, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

Often in rare diseases, families report insufficient information transmission after diagnosis and how this influences coping with the disease. In addition, perceptions of people living with a certain condition like CDG about the quality of the healthcare they receive are highly dependent on the quality of their interactions with their healthcare clinician and team. Actually, the connection that a patient and families feel with his or her clinician can ultimately improve their health mediated through participation in their care, adherence to treatment, and self-management for people living with a condition.

So far, no systematic study about information needs after the announcement of CDG diagnosis and people living with a condition/family – healthcare provider communication in CDG was done. An international research study named CDG Journey Mapping, is currently led by CDG & Allies PPAIN in collaboration with worldwide CDG patient groups, families and professionals.

During this session the collective results from the CDG Journey Mapping research study will be shared, including:

- Description of the journey faced by families and professionals when looking for information after announcement of CDG diagnosis.
- Where and how to look for reliable information about CDG?
- What's the role from patient groups in providing information and giving support after the announcement of CDG diagnosis?
- How to improve people's knowledge about CDG and enable access to reliable and relevant information?
- People living with CDG /families–physician communication: where do we stand? Major hurdles and solutions.

Day 1: Friday 14th May 2021 from 19:00 -19:15 PM, Lisbon, Portugal

Break

Keynote session Day 2, Friday 14th May 2021

Theme 3: Well-being and resilience skills for families and professionals.

Day 2: Friday
14th May 2021

19:15 – 20:45
PM, Lisbon,
Portugal

Introduction and session presentation by Moderator(s) (5 minutes):

Rita Francisco (Portugal), Vanessa Ferreira (Portugal) and Carlota Pascoal (Portugal)

Talk Title and corresponding Speaker(s) (15 minutes):

Building Resilience: Overcoming compassion, fatigue and burnout, by Vanessa Ferreira (CDG Portugal)

Round table discussion with Panelists facilitated by Moderator (s) (~35 minutes):

Rita Francisco (Portugal) Vanessa Ferreira (CDG Portugal).

Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021.

Panelists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Karen Morici (USA)
- Family and/or Patient Group Perspective: Mandy Pinheiro (South Africa)

- Family and/or Patient Group Perspective: Melissa Schlemmer (USA)
- Family and/or Patient Group Perspective: Karen Hackley (UK)
- Family and/or Patient Group Perspective: Yolandi Botha (South Africa)
- Family and/or Patient Group Perspective: Begoña Alonso Búa (Spain)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Clinician Perspective: Jaak Jaeken (Belgium)

Special Rare Disease Expert:

Nicola Miller, Rare Revolution Magazine (UK) (this expert will share learnings and best practices from another Rare Disease area that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~30 minutes):

Rita Francisco (Portugal) Vanessa Ferreira (CDG Portugal).

Moderators supported by: Marta Silva, Madalena Abade, Yasmin Pola, Ana Piedade and Salvador Magrinho from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

	Every day, people from all walks of life face health and personal challenges. A CDG diagnosis affects the entire support network, including family and healthcare providers. In this session, families and experts will discuss the importance of self-care and resilience, so that you can ultimately be the best care provider for your family. Several topics will be addressed including stories of resilience that offer hope and inspiration to others facing adversity.
--	---

- | | |
|--|---|
| | <ul style="list-style-type: none">● What is resilience? Why is resilience important when facing adversities and challenges?● Why might resilience be the most important thing you can do for yourself right now?● What are the 7Cs of resilience?● What are the essential skills that make you resilient?● Best practices on how to build and cultivate resilience● Tips to boost resilience when caregiving. Top self-care tips for CDG families and professionals● List of resources dedicated to resilience for CDG families and professionals |
|--|---|

20:45 – 21:15 PM Lisbon, Portugal: Virtual gather together.	
---	--

Day 3: Saturday 15th May 2021

Simultaneous interpretation today is offer in the following languages:

- English > Portuguese
- English> Italian
- English > Spanish
- English> French

Theme 4: CDG research and drug development: updates, challenges and solutions.

Saturday 15th May 2021 from 14:45 – 15:00 PM, Lisbon, Portugal

**Want to know what time corresponds in your country? Use World Time Buddy (WTB) at
<https://www.worldtimebuddy.com/>**

Virtual gathering to Secure you connect with some anticipation.

**Day 3: Saturday
15th May 2021**

**15:00 – 15:05
PM Lisbon,
Portugal**

Welcome to the 5th World Conference CDG by organisers.
Carlota Pascoal, Rita Francisco, Paula Videira and Vanessa Ferreira (Portugal).

Day 3: Saturday 15th May 2021 15:05 – 16:15 PM Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Rita Francisco (Portugal), Vanessa Ferreira (Portugal) and Carlota Pascoal (Portugal) Panel of discussion 3: CDG clinical research participation, education and awareness: Opportunities, challenges, and solutions from families and professionals' views. Talk Title and corresponding Speaker(s) 1 (15 minutes): Is Real-World Data and Real-World Evidence, the key to accelerate Congenital Disorders of Glycosylation (CDG) drug discovery and development?, by Alexandre Descalço Gil and Pedro Granjo Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal) Round table discussion with Panelists facilitated by Moderator (s) (15 minutes): Rita Francisco (Portugal) and Vanessa Ferreira (Portugal) Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos, Tiago Martins and Matilde Matos from Sci and Volunteer Program FCT NOVA 2021. Talk Title and corresponding Speaker(s) 2 (10 minutes): WorldCDG.Org, the web-based platform for educational and explanatory resources to empower the CDG community about Clinical Trials, by Yasmin Mei Pola (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal) Round table discussion with Panelists facilitated by Moderator (s) (15 minutes): Rita Francisco (Portugal) and Vanessa Ferreira (Portugal) Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos, Tiago Martins and Matilde Matos from Sci and Volunteer Program FCT NOVA 2021.
---	---

Panelists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Jenny Tupper (UK)
- Family and/or Patient Group Perspective: Nikki Zimmerman (USA)
- Family and/or Patient Group Perspective: Barbara Vulso (Ireland)
- Family and/or Patient Group Perspective: Ashleigh Linthicum (USA)
- Researcher Perspective: Giuseppina Andreotti (Italy)
- Clinician Perspective: Rita Barone (Italy)
- Clinician Perspective: Christina Lam (USA)
- Clinician Perspective: Stephanie Grunewald (UK)
- Clinician Perspective: Christin Johnsen (USA)
- Pharmaceutical industry Perspective: Helen Shapiro (Glycomine, USA)

Special Rare Disease Expert:

Daniel Lewi (this expert will share learnings and best practices from another Rare Disease area called Tay-Sachs that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~ 15 minutes):

- Attendees: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- Panelists & poster presenters: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

Clinical research has been fundamental in fostering the development of new treatments in medicine, and they are continually changing. The traditional model of evaluating treatments based primarily on primary outcome measures has stumbled in its application to rare disease. Rare disease clinical research faces the methodological challenges of small, heterogeneous populations and relatively few validated, disease-specific outcome measures. Although much progress has been made in developing scientific knowledge and conducting clinical research in rare diseases including CDG, serious challenges remain. During this session, the results from a survey administered within the CDG community will be shared as well as a successful CDG case study potentially transferable across countries and diseases. Thus, you will have the opportunity to listen about:

- What does involvement of people living with CDG and their family members within CDG clinical research mean to families and professionals?
- Why and how CDG families are contributing for clinical research?
- Which are the barriers faced by people living with CDG and their family members, when participating in clinical research?
- Identify solutions to increase participation in clinical research of people living with CDG and their family members.
- Which are the current main sources of information about existing clinical research on CDG?
- How aware about CDG clinical research are people living with CDG and family members?
- How and when to partner with people living with CDG, families and patient groups to increase clinical trial awareness, education and participation.

Day 3: Saturday 15th May 2021 from

16:15 – 16:30 PM, Lisbon, Portugal

BREAK

Day 3: Saturday 15th May 2021 16:30 – 18:00 PM Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Sandra Brasil (Portugal) and Vanessa Ferreira (Portugal) Panel of discussion 4: Overview of non-dietary supplementation approaches under development and current clinical trials for CDG. Talk Title and corresponding Speaker(s) 1 (18 minutes): Overview of non-dietary supplementation approaches under development and current clinical trials for CDG by Sandra Brasil (Portugal) Lightning to Posters session This is the chance for conference participants to listen about pioneering work in CDG. Poster presentation (s) (3 minutes): Epalrestat monotherapy in a Single Patient with Phosphomannomutase 2 Deficiency (PMM2-CDG) by Anna Natalia Ligezka (Mayo Clinic, USA) Round table discussion with Panelists facilitated by Moderator (s) (~40 minutes): Sandra Brasil (Portugal) and Vanessa Ferreira (Portugal) Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos, Tiago Martins and Matilde Matos from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. • Family and/or Patient Group Perspective: Holly Carmichael (USA) • Family and/or Patient Group Perspective: Yolanda Scott (Spain) • Family and/or Patient Group Perspective: Kara Berasi (USA)
--	--

- Researcher perspective: Belén Pérez González (Spain)
- Researcher perspective: Ethan Perlstein (USA)
- Researcher Perspective: Kristin Kantautas (Canada)
- Clinician Perspective: Eva Morava (USA)
- Clinician Perspective: Mercedes Serrano (Spain)
- Pharmaceutical industry Perspective: Horacio Plotkin (Glycomine, USA)
- Pharmaceutical industry Perspective: Shoshana Shendelman (Applied Therapeutics, USA)
- Pharmaceutical industry Perspective: Jahannaz Dastgir (Applied Therapeutics, USA)

Special Rare Disease Expert:

Marisol Montolio (this expert will share learnings and best practices from another Rare Disease area named Duchenne Muscular Dystrophy that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~20 minutes):

Sandra Brasil (Portugal) and Vanessa Ferreira (Portugal)

Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos, Tiago Martins and Matilde Matos from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees:** please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters:** please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

	Session Brief description: Congenital disorders of glycosylation (CDG) are a group of clinically and genetically heterogeneous metabolic disorders. Over approximately 170 CDG types have been described. Most CDG types are ultrarare disorders. Families and professionals are united to make therapies a short-term reality for people living with CDG. This session will walk you throughout: • Overview of currently recruiting, ongoing and upcoming clinical trials and research studies for CDG, including: Natural History studies and Interventional Clinical Trials (several approaches will be addressed such as transplantation, gene therapy, liposomes, antisense therapy, drug repurposing - notably, acetazolamide, epalrestat- and pharmacological chaperones). • Identify major challenges faced when developing non-dietary supplementation approaches and trials for CDG. • Identify solutions to boost development of non-dietary therapeutic approaches and trials for CDG. • Lessons from other rare diseases than can be transferred for CDG and help to faster therapeutic approaches. • Final recommendations to faster non-dietary therapeutic approaches and trials for CDG.
Day 3: Saturday 15th May 2021 from 18:00 – 18:15 PM, Lisbon, Portugal BREAK	
Day 3: Saturday 15th May 2021	Introduction and session presentation by Moderator(s) (5 minutes): Sandra Brasil (Portugal), Begoña Cano (Spain) and Rita Francisco (Portugal) Panel of discussion 5: Update on ongoing and under investigation dietary supplementation therapies in Congenital Disorders of Glycosylation.

18:15 – 19:45 PM, Lisbon, Portugal	Talk Title and corresponding Speaker(s) 1 (18 minutes): Update on ongoing and under investigation dietary supplementation therapies in Congenital Disorders of Glycosylation, by Sandra Brasil (Portugal) Lightning to Posters session This is the chance for conference participants to listen about pioneering work in CDG. Poster presentation (s) (3 minutes): Sugar-bisphosphates to cure PMM2-CDG? by Mariateresa Allocca (Institute of Biomolecular Chemistry - National Research Council (ICB-CNR), Italy) Round table discussion with Panelists facilitated by Moderator (s) (~40 minutes): Sandra Brasil (Portugal), Begoña Cano (Spain) and Rita Francisco (Portugal) Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos and Tiago Martins from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. • Family and/or Patient Group Perspective: Hagint Babakhanian (USA) • Family and/or Patient Group Perspective: Claudia Vasquez (USA) • Family and/or Patient Group Perspective: Larissa Fonseca Andrade Vieira (Brasil) • Family and/or Patient Group Perspective: Begoña Cano (Spain) • Family and/or Patient Group Perspective: Gabriel Ribeiro (Portugal) • Researcher perspective: Christian Thiel (Germany) • Clinician Perspective: Peter Witters (Belgium) • Clinician Perspective: Eva Morava (USA)
--	---

- Pharmaceutical industry Perspective: Jeff Wilkins (Cerecor, USA)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~20 minutes):

Sandra Brasil (Portugal), Begoña Cano (Spain) and Rita Francisco (Portugal).

Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos and Tiago Martins from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees:** please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters:** please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Short Brief description:

Mannose-Phosphate Isomerase (MPI)-CDG was the first treatable CDG by high dose mannose supplements. Recently, with the successful use of d-galactose in Phosphoglucomutase 1 (PGM1)-CDG, other CDG types have been tested on galactose and other sugars. In addition, there is an increasing potential of dietary supplementation therapies for CDG. During this session, an update as for ongoing and under investigation dietary supplementation therapies and future perspectives:

- Where do we stand as for supplementation with sugars (e.g., mannose, galactose, fucose), nucleotides (e.g., uridine) and trace elements (e.g., manganese, magnesium) in CDG.
- Where do we stand as for Biotin research study and CDG?
- Identification of challenges faced when setting up dietary supplementation therapies for CDG.
- Assess solutions to continue developing dietary supplementation therapies for CDG.

	• Future perspectives on dietary supplementation therapies for CDG.
Day 3: Saturday 15 th May 2021 from 19:45 – 20:00 pm, Lisbon, Portugal	
BREAK	
Theme 5: Tools to make CDG therapies an approved reality!	
Day 3: Saturday 15 th May 2021 20:00 – 21:30 PM, Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Luísa Barros (Portugal), Carlota Pascoal (Portugal), Inês Ferreira (Portugal) and Catarina Teixeira (Portugal) and Eduardo Eliziário de Almeida (Portugal). Community CDG Think Tank 3: Clinical Outcome Assessments (COAs): Where do we stand as for COAs for CDG? Talk Title and corresponding Speaker(s) 1 (18 minutes): Patient reported outcome measures to enable PMM2-CDG symptom-specific quality of life assessment: listening to what matters from the point of view of people living with CDG, their family members and professionals', by Eduardo Eliziário de Almeida (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal). Lightning to Posters session This is the chance for conference participants to listen about pioneering work in CDG. Poster presentation (s) (3 minutes):

	Evaluating Association of Nijmegen Paediatric CDG Rating Scale (NPCRS) with Patient Reported Outcome Measurement Information System (PROMIS) in Patients with Phosphomannomutase 2 Deficiency (PMM2-CDG) by Anab Mohamed (Mayo Clinic, USA) Round table discussion with Panelists facilitated by Moderator (s) (40 minutes): Luísa Barros (Portugal), Carlota Pascoal (Portugal), Inês Ferreira (Portugal) and Catarina Teixeira (Portugal) and Eduardo Eliziário de Almeida (Portugal). Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos and Tiago Martins from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. ● Family and/or Patient Group Perspective: Stacey Vogelee (USA) ● Family and/or Patient Group Perspective: Jody Goldhawk (Canada) ● Family and/or Patient Group Perspective: Lorenzo Lachi (Italy) ● Family and/or Patient Group Perspective: Konstantin Feinberg (Canada) ● Family and/or Patient Group Perspective: Marta Vazquez Gómez (Spain) ● Researcher Perspective: Elodie Lebrondochel (France) ● Clinician Perspective: Andrew C. Edmondson (USA) ● Clinician Perspective: Dafne Horowitz (Brasil) ● Clinician Perspective: Eva Morava (USA) ● Pharmaceutical industry Perspective: Alison Slade (UK) ● Pharmaceutical industry Perspective: John Boland (Cerecor, USA) Special Rare Disease Expert:
--	--

Luísa Barros (Portugal) (this expert will share learnings and best practices from another Rare Disease area that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~20 minutes):

Luísa Barros (Portugal), Carlota Pascoal (Portugal), Inês Ferreira (Portugal) and Catarina Teixeira (Portugal) and Eduardo Eliziário de Almeida (Portugal).

Moderators supported by: Marta Falcao, Pedro Granjo, Inês Santos and Tiago Martins from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

Clinical Outcome Assessments (COAs) is a measure that describes or reflects how a patient feels, functions, or survives. COAs including patient-reported outcome (PRO) measures, are routinely used in drug development and other clinical research initiatives. COAs are an essential component of clinical trials to collect stakeholders perspectives on treatment benefits and risks. Thus, it is crucial during medical product development to incorporate the perspective from people living with a rare disease and their family members in rare disease trials. Understanding the value of these treatments to the people living with a certain condition is increasingly important to help payers and providers, as well as people with a condition, to make good medical decisions. Outcomes research in rare disease clinical trials has been hampered by diverse measurement challenges due to the small, heterogeneous, and widely dispersed nature of typical rare disease populations. Currently, there is not a COA specific for CDG.

	As COAs are a critical component of treatment development, back to 2018 the CDG & Allies PPAIN, started to build a roadmap to successfully build and implement COAs within CDG clinical research. For the first time, the results from the development of a Clinical Outcome Assessments (COA) tool for CDG, will be shared and discussed, including: • Why, what and How to best understand the CDG disease or condition? • Why, what and How to conceptualize treatment benefits in CDG? • Why, what and How to assess existing outcome measurement potentially transferable for CDG? • Why, what and How to select/build a CDG-COA? • Challenges and solutions faced when building a CDG-COA. • Overview of best practice to build CDG-COAs applicable across rare diseases.
--	---

Day 4: Sunday 16th May 2021 & World CDG Awareness day 2021

Simultaneous interpretation today is offer in the following languages:

- English > Portuguese
- English> Italian
- English > Spanish
- English> French

Theme 6: How new technologies and tools can boost CDG basic research and therapies.

Sunday 16th May 2021 from 12:30 -13:00 PM Lisbon, Portugal

**Want to know what time corresponds in your country? Use World Time Buddy (WTB) at
<https://www.worldtimebuddy.com/>**

Virtual gathering to Secure you connect with some anticipation.

**Day 4: Sunday 16th
May 2021**

**13:00 -13:05 PM
Lisbon, Portugal**

Welcome to the 5th World Conference CDG by organisers.

Carlota Pascoal, Rita Francisco, Paula Videira and Vanessa Ferreira (Portugal).

Day 4: Sunday 16th May 2021 13:05 -14:00 PM Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Vanessa Ferreira (Portugal), Gonalo Valado (Portugal) and Tatiana Rijoff (Switzerland) Panel of discussion 6: Artificial Intelligence (AI), Bioinformatics and Multi-Omics approaches to encourage potential therapies for CDG. Talk Title and corresponding Speaker(s) 1 (~15 minutes) Artificial Intelligence (AI), Bioinformatics and Multi-Omics approaches to encourage potential therapies for CDG, by Gonalo Valado (Portugal) and Tatiana Rijoff (Switzerland) Round table discussion with Panelists facilitated by Moderator (s) (~10 minutes): Vanessa Ferreira (Portugal), Gonalo Valado (Portugal) and Tatiana Rijoff (Switzerland) Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. ● Researcher Perspective: Sandra Brasil (Portugal) ● Researcher Perspective: Gonalo Valado (Portugal) ● Researcher Perspective: Tatiana Rijoff (Switzerland) ● Researcher Perspective: Silvia Bottini (France) ● Researcher Perspective: Jan Mucha (Slovakia) ● Clinician Perspective: David Coman (Australia) ● Pharmaceutical industry Perspective: Teresa Sardon (Anaxomics, Spain)
---	--

Lightning to Posters session

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes per presentation):

- Multi-omics approaches to improve rare disease diagnosis: challenges, advances and perspectives by Justine Labory (Université Côte d'Azur, France)
- Developing new technologies for diagnostics of congenital disorders of glycosylation, by Merel Post (Radboud University Medical Center, The Netherlands)
- Relative Quantification of Glycans as a Diagnostic Approach of Congenital Disorders of Glycosylation, by Jaime Moritz Brum (Brazil)
- Where we stand as for CDG research in Slovakia by Jan Mucha (Slovakia)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~12 minutes):

Vanessa Ferreira (Portugal), Gonçalo Valadão (Portugal) and Tatiana Rijoff (Switzerland)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees:** please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters:** please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

	The development of new technologies, such as genomic analysis by means of next generation sequencing (NGS) and other “omics technologies”, has boosted the molecular understanding and diagnosis of RDs. However, there is a growing need to develop new methods to integrate multi-omics data from different technologies. Furthermore, the ability of AI technologies to integrate and analyze data from different sources (e.g., multi-omics, registries) can be used to overcome further challenges, such as low diagnostic rates, reduced number of people living with a condition, and geographical dispersion. Ultimately AI-mediated knowledge could significantly boost therapy development for RDs like CDG. These approaches together can solve RDs challenges such as low diagnostic rates, reduced number of people living with a certain rare disease, geographical dispersion, limited annotated data. During this session, we will walk you throughout: • The application of AI, Bioinformatics and Multi-Omics approaches in RD, notably in CDG. • Sum up of tools used or with potential to be used in CDG from diagnosis to therapies. • Key challenges, advances and perspectives when using AI, Bioinformatics and Multi-Omics approaches in RD, notably in CDG. • Recommendations to boost use of these new technologies in the field of CDG
14:00 – 14:15 PM, Lisbon, Portugal	BREAK
Theme 7: CDG child, teen and adult care and management	
Day 4: Sunday 16 th May 2021 14:15 – 15:15 PM, Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Vanessa Ferreira (Portugal), Carlota Pascoal (Portugal) and Rita Francisco (Portugal)

Panel of discussion 7: Standards of care and management for CDG children, teens and adults: Identify challenges and solutions to promote holistic and patient-centred care planning and care coordination across countries.

Talk Title and corresponding Speaker(s) 1 (10 minutes):

A grassroots effort to build community practical tools for CDG: Spotlight to PMM2-CDG international guidelines a model transferable among all CDG types, by Susana Alves (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal).

Lightning to Posters session

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes):

- Neurological manifestations in PMM2 related congenital disorders of glycosylation (CDG): Insights into clinico-radiological characteristics and recommendations for follow-up by Karthik Muthusamy (Mayo Clinic, USA)

Round table discussion with Panelists facilitated by Moderator (s) (~10 minutes):

Vanessa Ferreira (Portugal) and Rita Francisco (Portugal)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

Panelists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Oriane Moreau (France)
- Family and/or Patient Group Perspective: Sandra Pereira Pinto (Spain & Portugal)
- Family and/or Patient Group Perspective: Louise Rimen (Denmark)
- Family and/or Patient Group Perspective: Alicia Fazakas (USA)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Clinician Perspective: Ruqaiyah Altassan (Saudi Arabia)
- Clinician Perspective: Donna Krasnewich (USA)
- Pharmaceutical industry Perspective: Jeff Wilkins (Cerecor, USA)

Talk Title and corresponding Speaker(s) 2 (8 minutes):

Transforming the International consensus guidelines for phosphoglucomutase 1 deficiency (PGM1-CDG) into readable and understandable resources: a model transferable across all CDG types, Matilde Matos (Science and Volunteer program, NOVA School of Science and CDG & Allies PPAIN, Portugal).

Lightning to Posters session

This is the chance for conference participants to listen about pioneering work in CDG.

Poster presentation (s) (3 minutes):

- Successful heart transplantation in a PGM1-CDG infant by Ruqaiyah Altassan (Saudi Arabia)

Round table discussion with Panelists facilitated by Moderator (s) (~15 minutes):

Vanessa Ferreira (Portugal) and Rita Francisco (Portugal)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

Panelists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Oriane Moreau (France)
- Family and/or Patient Group Perspective: Sandra Pereira Pinto (Spain & Portugal)
- Family and/or Patient Group Perspective: Louise Rimen (Denmark)
- Family and/or Patient Group Perspective: Alicia Fazakas (USA)
- Clinician Perspective: Ruqaiyah Altassan (Saudi Arabia)
- Clinician Perspective: Donna Krasnewich (USA)
- Pharmaceutical industry Perspective: Jeff Wilkins (Cerecor, USA)

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~10 minutes):

Vanessa Ferreira (Portugal) and Rita Francisco (Portugal)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees:** please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters:** please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

	During this session, we wish to promote ideas and measures that facilitate multidisciplinary, holistic, continuous, person-centred and participative care provision to people living with CDG. We will discuss and exchange experiences and views about: • Potential challenges and solutions in implementing CDG standards of care across countries. • Exchange ideas to simplify care pathways & support navigation for people living with CDG, their families and professionals • Assess current support transition periods to prevent care ruptures and facilitate independent life • Determine how to create the link/facilitate coordination between services & professionals. • Set up a care and life plan with a person with CDG and their family: major challenges and solutions. • Define short- and long-term strategy to ensure CDG holistic and people-centred care planning and care coordination.
15:15 – 15:30 PM, Lisbon, Portugal	BREAK
Theme 8: The impact of COVID-19 on CDG, as well as the opportunities embraced, notably the use of digital health to improve diagnosis, treatment, navigation and care coordination, and integration and coordination for broader societal and wellbeing of people living with a certain condition.	
Day 4: Sunday 16th May 2021 15: 30 – 17:00 PM, Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Vanessa Ferreira (Portugal), Alexandre Gil (Portugal), Carlota Pascoal (Portugal) and Madalena Parrado (Portugal).

Panel of discussion 8: How COVID-19 impacted the life and care of people living with CDG and their caregivers? Was COVID-19 the booster for the use of digital health among the CDG community?

Talk Title and corresponding Speaker(s) 1 (15 minutes):

How COVID-19 impacted the life and care of people living with rare diseases like CDG and their caregivers? Was COVID-19 the booster for the use of digital health among the CDG community? by Alexandre Gil (Portugal).

Round table discussion with Panelists facilitated by Moderator (s) (45 minutes):

Vanessa Ferreira (Portugal), Alexandre Gil (Portugal), Carlota Pascoal (Portugal) and Madalena Parrado (Portugal).

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

Panelists & Special Rare Disease Expert:

Poster presenters are also invited to share views during the round table.

Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared.

- Family and/or Patient Group Perspective: Fiona Waddell (The Netherlands)
- Family and/or Patient Group Perspective: Marit Kuyper (The Netherlands)
- Researcher Perspective: Jaime Brum (Brasil)
- Researcher Perspective: François Foulquier (France)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Clinician Perspective: Saadet Mercimek-Andrews (Canada)
- Clinician Perspective: Andrew C. Edmondson (USA)
- Pharma Perspective: Peter Williams (Glycomine, USA)

Special Rare Disease Expert:

Begoña Nafría (Spain) (this expert will share learnings and best practices from other Rare Diseases that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (20 minutes):

Vanessa Ferreira (Portugal), Carlota Pascoal (Portugal) and Madalena Parrado (Portugal)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees**: please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters**: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

COVID-19 has created profound challenges for drug developers, health care professionals, researchers, people living with a condition and carers to overcome. It has forced everyone in the rare disease world to think and work using new and innovative ways, such as digital health. Digital health is a broad category encompassing electronic health, mobile health, telehealth and health data, among others. It offers solutions that can strengthen health systems, such as bringing health services directly to people's homes and to underserved communities, helping to map outbreaks of disease, and integrating digital tools that make healthcare more responsive and productive.

During this panel we will:

- Identify the impact of COVID-19 on CDG daily lives and care of people living with CDG and their carers
- Identify the impact of COVID-19 on CDG drug development and access to medicines

	• Assess strategies of resilience from the CDG community at different levels during COVID-19 pandemic • Collect lessons from COVID-19 to ensure appropriate care for CDG in case of new health emergencies • So, what is digital health? Why digital health and rare diseases? • What's happening in digital health for rare diseases, notably for CDG? • Which digital health initiatives can be transferred for CDG? • Identify challenges and opportunities for digital health in CDG. • How can we speed up the use of digital health in CDG without risks? • Final recommendations as for the use of digital health to strength CDG care after COVID-19 times.
17:00 – 17:30 PM Lisbon, Portugal	BREAK
Day 4: Sunday 16th May 2021 17:30 – 17: 45 PM, Lisbon, Portugal	World CDG Awareness day 2021 celebrations Let's unravel UNITED the #CDGMosaic May 16, 2021, during the World CDG Awareness day celebrations. Read more <u>HERE</u> The #CDGMosaic honors all children and adults that live with CDG and their family members.

“Love is the essence of all we do. CDG families will never give up! And we have genuine professionals supporting us. Thus, just do it and keep going! By Vanessa Ferreira.



Theme 9: World CDG Community – Why, What and How from stakeholders views and experiences.

**Day 4: Sunday 16th
May 2021**

Introduction and session presentation by Moderator(s) (5 minutes):
Eleonora Passeri, Vanessa Ferreira (Portugal), Rita Francisco (Portugal) and Madalena Parrado (Portugal)

17:45 – 19:30 PM Lisbon, Portugal (a break will be done)	Panel of discussion 9: How to best serve the CDG community across countries? Key challenges and solutions by stakeholders views. Talk Title and corresponding Speaker(s) 1 (15 minutes) : How to best serve the CDG community across countries? Key challenges and solutions by stakeholders views, by Vanessa Ferreira (Portugal) Lightning to Posters session This is the chance for conference participants to listen about pioneering work in CDG. Poster presentation (s) (3 minutes): Clinical case with a new type CDG-Ix from Bulgaria, Malina Stancheva-Ivanova (Bulgaria). Round table discussion with Panelists facilitated by Moderator (s) (~45 minutes): Eleonora Passeri, Vanessa Ferreira (Portugal), Rita Francisco (Portugal) and Madalena Parrado (Portugal) Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. • Family and/or Patient Group Perspective: Tata Tsintsadze (CDG Georgia) • Family and/or Patient Group Perspective: Juliana da Silva Ferreira (Brasil) • Family and/or Patient Group Perspective: Etienne Barrier (Estonia) • Family and/or Patient Group Perspective: Adamastor Kemmler (Brasil) • Family and/or Patient Group Perspective: Paul Collot (México)
--	---

- Family and/or Patient Group Perspective: Kulani Shiluvane (South Africa)
- Family and or Patient Group Perspective: Nathalie Harvey (France)
- Researcher Perspective: Marina Szlago (Argentina)
- Researcher Perspective: Iván Martínez-Duncker (México)
- Researcher Perspective: Carla G. Asteggiano (Argentina)
- Researcher Perspective: Dulce Quelhas (Portugal)
- Researcher Perspective: Ida Vanessa Doederlein Schwartz (Brasil)
- Clinician Perspective: Charles Marques Lourenço (Brasil)
- Clinician Perspective: Malina Stancheva-Ivanova (Bulgaria)
- Pharma Perspective: Horacio Plotkin (Glycomine,USA)
- Pharma Perspective: Dottie Caplan (Applied Therapeutics)

Special Rare Disease Expert:

Eleonora Passeri (this expert will share learnings and best practices from other Rare Diseases that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (30 minutes):

Eleonora Passeri, Vanessa Ferreira (Portugal), Rita Francisco (Portugal) and Madalena Parrado (Portugal)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees:** please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.

	● Panelists & poster presenters: please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters. ● Panelists and poster presenters are strongly encouraged to pose questions and share views and experience. Session Brief description: In the previous 4th World CDG Congress held in 2019, the CDG & Allies PPAIN in collaboration with worldwide families and professionals, began the very important endeavour of systematically assessing the needs of the CDG community regarding drug development. In preparation for the upcoming 5th World CDG Congress (13-16 May 2021), CDG & Allies PPAIN will launch an online survey aimed at evaluating information-seeking behaviours and educational needs and preferences. Such information is needed to inform clinical guidelines, educational initiatives and new models of healthcare for children and adults with rare diseases. Together we will, ● Where do we stand as for interest in finding and utilising resources and services from patient groups? ● What are the five most important needs from our community across countries and stakeholders? ● Why are these needs the most important? What factors may contribute to these community needs? ● What resources already exist to address the identified community needs? ● What resources are still needed to address the identified community needs? ● Are any of the identified community needs already being addressed? ● Select priority CDG Community actions to best serve the CDG community across countries. ● Define how to measure impact and progress of the implementation of priority CDG Community actions to best serve the CDG community across countries. ● How can we continue to engage the CDG community across countries? ● What is the best communication strategy to ensure coordination across countries and stakeholders?
19:30 – 19.45 PM, Lisbon, Portugal	BREAK

Closing plenary session Day 4 16 May 2021 (Sunday, World Awareness Day for CDG)	
Day 4: Sunday 16th May 2021 19:45 – 20: 45 PM, Lisbon, Portugal	Introduction and session presentation by Moderator(s) (5 minutes): Eleonora Passeri, Vanessa Ferreira (Portugal), Rita Francisco (Portugal) and Madalena Parrado (Portugal) Keynote speech: Blue sky is the limit: looking ahead for CDG. Round table discussion with Panelists facilitated by Moderator (s) (~45 minutes): Eleonora Passeri , Vanessa Ferreira (Portugal), Rita Francisco (Portugal) and Madalena Parrado (Portugal) Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021. Panelists & Special Rare Disease Expert: Poster presenters are also invited to share views during the round table. Some experiences and views when living with CDG (or when working in the field of CDG/rare diseases) will be shared. • Family and/or Patient Group Perspective: Emma Finklaire (Australia) • Family and/or Patient Group Perspective: Andrea Miller (USA) • Family and or Patient Group Perspective: Kerry Blondheim (USA) • Researcher Perspective: Paula Videira (Portugal) • Researcher Perspective: Chema Fernández (Spain) • Clinician Perspective: Jaak Jaeken (Belgium)

- Clinician Perspective: Erik Eklund (Sweden)
- Clinician Perspective: Marc Patterson (USA)
- Clinician Perspective: Mercedes Serrano (Spain)

Special Rare Disease Expert:

Eleonora Passeri (this expert will share learnings and best practices from another Rare Disease area that can be transferred for CDG families and professionals across countries).

Open virtual floor for Questions and Answers facilitated by Moderator(s) (~15 minutes):

Eleonora Passeri, Vanessa Ferreira (Portugal), Rita Francisco (Portugal) and Madalena Parrado (Portugal)

Moderators supported by: Marta Falcao, Madalena Abade, Joana Grilo, Tiago Martins and Madalena Raposo from Sci and Volunteer Program FCT NOVA 2021.

- **Attendees:** please use the Questions and Answers (Q&A) feature from ZOOM. We strongly encourage Questions and Answers from all participants about firstly the poster presentation(s) (when applicable), and then followed by questions targeting what was listened during the session.
- **Panelists & poster presenters:** please use the Chat to post your questions and they will be transferred to the Q&A by the moderators' supporters.
- Panelists and poster presenters are strongly encouraged to pose questions and share views and experience.

Session Brief description:

A prestigious panel will help us wrap up our 5th World Conference on CDG for families and professionals with a look ahead at 2022.

Day 4: Sunday 16th May 2021 20:30 – 20:45 PM, Lisbon, Portugal	Final conclusions, next steps and Wrap -up
--	---

World CDG Awareness day 2021 celebrations

We will unravel the #CDGMosaic May 16, 2021, during the World CDG Awareness day celebrations.

Read more [HERE](#)