

4TH WORLD CONFERENCE ON CDG | 2019

FOR FAMILIES AND PROFESSIONALS

26 / 27TH JULY 2019

LISBON (PORTUGAL)

1ST CDG SATELLITE
MEETING | 2019

FOR PROFESSIONALS

25TH JULY 2019

LISBON (PORTUGAL)



THE CDG ROAD

FROM DIAGNOSIS
TO THERAPIES



Congenital Disorders
of Glycosylation

Final Agenda

DAY 1 | 25th JULY 2019

What: CDG Satellite Meeting for Professionals.

Where: Room 6 of Faculty of Psychology of University of Lisbon.

Hours	Session Topic	Theme	Title/ Speaker
13.30 - 14.00	Registration		
14.00 - 14.30	CDG Nomenclature	What is a CDG?	State of the art for CDG Nomenclature Carlos Ferreira INIH, USA
14.30 - 16.30			<p>Discussion session: The Need for Unification: Challenges of CDG Nomenclature and Classification.</p> <p>Moderators: Eva Morava, Jaak Jaeken, Maurizio Scarpa, Stephanie Grunewald, Paula Videira.</p> <p>Observers: CDG & Allies PPAIN researchers and selected volunteers (deliverable: aligned notes from session).</p>
16.30 - 17.00	Coffee break		
17.00 - 18.30	CDG world collaborations to accelerate basic research and therapeutic development	How to reinforce collaborative work? Identification of existing major barriers. Set up plan of action to accelerate world collaborations.	<p>Pinpoint existing initiatives and Spotlight existing ones:</p> <ul style="list-style-type: none"> • CDG & Allies Professionals and Patient Associations International Network (CDG & Allies PPAIN); • A new CDG consortium; • MetabERN (European Reference Network for Rare Hereditary Metabolic Diseases); • CDG Spanish-Consortium; • CDG Australian network. <p>Moderators: Eva Morava, Jaak Jaeken, Maurizio Scarpa, Marc Patterson, Mercedes Serrano, David Coman and CDG & Allies PPAIN researcher (name TBC).</p> <p>Observers: CDG & Allies PPAIN researchers and selected volunteers (deliverable: aligned notes from session).</p>
19.00	United to find a restaurant and have dinner together and continue fruitful discussions.		

DAY 2 | 26th JULY 2019

What: World Conference on CDG for Families and Professionals.

Where: Auditorium Prof Simões dos Santos – Faculty of Dental Medicine of University of Lisbon.

Hours	Session Topic	Theme	Title/ Speaker
08.00 - 09.30	Registration and Poster Viewing		
09.30 - 10.00	<p>Welcome session CDG & Allies Professionals and Patient Associations International Network (CDG & Allies PPAIN) (on behalf of Organizing committee) Where do we stand as for Drug development and life cycle for CDG? Setting the pillars for 2 days conference!</p>		



WORKSHOP 1: Research and development

Hours	Session Topic	Theme	Title/ Speaker
10.00 - 10.15	Biobanks and Biomarkers in Rare Diseases		Biomarkers as invaluable tool for CDG diagnosis and therapeutic advances. By Stephanie Grunewald Great Ormond Street Hospital (GOSH), UK.
10.15 - 10.25			Where do we stand as for CDG Biobanks and Biomarkers at International level? Results from the 4th World Conference CDG Survey. By CDG & Allies - PPAIN.
10.25 - 11.05	Think Tank 1	<p>Keynote Moderator: Prof. Jaak Jaeken. Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Christina Lam, Paula Videira, Dulce Quelhas, Stephanie Grunewald.</p> <ul style="list-style-type: none"> • What are the 4 main obstacles to CDG Biobanks and Biomarkers? (10') • What are the 4 main solutions to CDG Biobanks and Biomarkers? (10') <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 3 main obstacles and priorities to address CDG Biobanks and Biomarkers and devise a short term plan of action. (20'). 	
11.05 - 11.35	Coffee break and Poster viewing and presentation		

WORKSHOP 2: Tools for pre-clinical and clinical drug development and approval

Hours	Session Topic	Theme	Title/ Speaker
11.35 - 11.50	Patient Registries (PRs)		Case study 1: A European patient registry for rare metabolic disease. By Maurizio Scarpa MetabERN, IT.
11.50 - 12.00			Case study 2: CDG Connect. By Holly Carmichael CDG CARE, USA.
12.00 - 12.15	Natural History Studies (NHS)		Case study 1: CDG Natural History Study sponsored by Glycomine. By Eva Morava Mayo Clinic, USA.
12.15 - 12.25			Case study 2: NIH CDG Natural History study. By Lynne Wolfe NIH, USA.
12.25 - 12.35			Where do we stand as for CDG PRs and NHS at International level? Results from the 4th World Conference CDG Survey. By CDG & Allies - PPAIN.
12.35 - 13.15	Think Tank 2	<p>Keynote Moderator: Prof. Marc Patterson Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Lynne Wolfe, Peter Witters, Eva Morava.</p> <ul style="list-style-type: none"> • What are the 4 main obstacles to CDG Patient Registries and Natural History Studies (NHS)? (10') • What are the 4 main solutions to CDG Patient Registries and Natural History Studies (NHS)? (10') <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 3 main obstacles and priorities to address CDG Patient Registries and Natural History Studies (NHS) and devise a short term plan of action. (20'). 	
13.15 - 15.15	Lunch and Poster viewing		

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WORKSHOP 3: Emerging tools/methods to accelerate therapeutic discovery

Hours	Session Topic	Theme	Title/ Speaker
14.30 - 14.40	Systems Biology for therapeutic discovery		How can Systems Biology can boost therapies for CDG and related diseases? By Teresa Sardon Anaxomics, ES.
14.40 - 14.55	Artificial Intelligence (AI)		Artificial Intelligence (AI): an introduction. By Gonçalo Valadão Autónoma University Lisbon, PT.
14.55 - 15.05			Case study 1: Artificial intelligence at the service of CDG diagnosis: Early predictive dysmorphic features of PMM2-CDG. By Antonio Martinez-Monseny Hospital Sant Joan de Déu, ES.
15.05 - 15.15			Case study 2: Application of AI in diagnostics of GPI biosynthesis defects: advances and pitfalls. By Alejex Knaus Institute for Genomic Statistics and Bioinformatics, GE.
15.15 - 15.30			Drug repositioning to accelerate drug development for Rare Diseases. (Remote presence). By Rick Thompson Findacure, UK.
15.30 - 15.45	Metabolomics		How can Metabolomics accelerate CDG and related diseases therapies? Antonio Pineda Hospital Universitari i Politècnic la Fe, ES.
15.45 - 16.25	Think Tank 3	<p>Keynote Moderator: Prof. Tamaz Kozicz Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Teresa Sardon, Gonçalo Valadão, Antonio Martinez-Monseny, Antonio Pineda.</p> <ul style="list-style-type: none"> • What are the 4 main challenges/obstacles for further implementing and developing these tools in biomedicine? (10'). • How could these emerging tools/methods accelerate therapeutic discovery? What are the 4 main solutions/areas of impact that these tools can promote?(10'). <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will reach consensus as for the 3 main obstacles and opportunities to address these new techniques for CDG and devise a short term plan of action. (20'). 	
16.25 - 16.45	Coffee break and Poster viewing and presentation		

WORKSHOP 4: CDG models

16.45 - 17.00	In vitro models	Modelling diseases process in dish: Induced pluripotent stem cells (iPSCs) technology. By Catarina Brito ITQB, PT.
17.00 - 17.10		The yeast and CRISPR/Cas to help therapeutic discovery on CDG. By François Foulquier Université des Sciences et Technologies, FR.
17.10 - 17.25	In vivo model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research	Zebrafish model for PMM2-CDG. By Paola de Haas Radboudumc, NL.
17.25 - 17.35		Mouse model for PMM2-CDG. By Andrew Edmondson Children's Hospital of Philadelphia, USA.
17.35 - 17.50		Where do we stand as for CDG disease models at International level? Results from the 4th World Conference CDG Survey. By CDG & Allies - PPAIN.



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WORKSHOP 4: CDG models

Hours	Session Topic	Theme	Title/ Speaker
17.50 - 18.30	Think Tank 4	<p>Keynote Moderator: Dr. Mercedes Serrano Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Ethan Perlstein, François Foulquier, Paola de Haas, Andrew Edmondson.</p> <ul style="list-style-type: none"> • What are the 4 main obstacles to the development/use of in vitro and in vivo model organisms in the field of CDG? (10’). • What are the 4 main priorities to the development/use in vitro and in vivo model organisms in the field of CDG? (10’). <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 3 main obstacles and priorities to address in vitro and in vivo model organisms in the field of CDG and devise a short term plan of action. (20’). 	
18.30 - 19.00	Break (time to talk and network)		
19.00 - 19.40	Surprise for families and professionals		
19.40 - 22.00	Dinner & Social Event (all together)		

DAY 3 | 27th JULY 2019

What: World Conference on CDG for Families and Professionals.

Where: Auditorium Prof Simões dos Santos – Faculty of Dental Medicine of University of Lisbon.

8.00 - 9.00	Gather together and Poster viewing		
WORKSHOP 5: Drug approval and access to patients			
9.00 - 9.20	Regulatory approval & Market access	Drug approval and market access: Can patients make a difference? By Alastair Kent UK.	
9.20 - 9.30		How did Cerecor do it? The case study of getting FDA approval for CDG therapy. By Perry Calias Cerecor Inc., USA.	
9.30 - 9.40		How the patient community can be empowered to get involved in drug development. By Inês Alves PT.	
9.40 - 9.50		Where do we stand for drug access and approval for CDG? Results from the 4th World Conference CDG Survey. By CDG & Allies - PPAIN.	
9.50 - 10.30	Think Tank 5	<p>Keynote Moderator: Alastair Kent Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Esmeralda Martins, Perry Calias, Inês Alves, Anabela Bandeira.</p> <ul style="list-style-type: none"> • What are the 4 main obstacles for regulatory approval in the field of CDG? (10’). • What are the 4 main priorities for regulatory approval in the field of CDG? (10’). <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 3 main obstacles and priorities to address regarding regulatory approval & market access in the field of CDG and devise a short term plan of action. (20’). 	
10.30 - 11.00	Coffee break and Poster viewing		

DAY 3 | 27th JULY 2019

What: World Conference on CDG for Families and Professionals.

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WORKSHOP 6: CDG clinical impact

Hours	Session Topic	Theme	Title/ Speaker
11.00 - 11.10	CDG Diagnosis		Challenges in CDG diagnosis. By Dorinda Marques-da-Silva APCDG, PT.
11.10 - 11.20			Opportunities for CDG diagnostics. By Dulce Quelhas Centro Hospitalar Universitário do Porto.
11.20 - 11.30	Clinical impact and outcomes		Genetic modifiers for PMM2-CDG diagnostics. By Giuseppina Andreotti Istituto di Chimica Biomolecolare, IT.
11.30 - 11.40			Movement disorders as clinical features of CDG. By Rita Barone UNICT, IT.
11.40 - 11.50			ImmunoCDGQ results. By Rita Francisco UCIBIO, CDG & Allies - PPAIN, PT.
11.50 - 12.05			PMM2-CDG guidelines and standards of care. By Ruqaiyah Altassan McGill University Health Centre, CA.
12.05 - 12.15			Outcome assessment tools for rare diseases. By Carlota Pascoal CDG & Allies PPAIN, PT.
12.15 - 12.25			How CDG impacts patients and family members. Sum up of coping strategies in CDG families. By Luísa Barros University of Lisbon, PT.
12.25 - 12.35			What do we know about and where do we stand for QoL and clinical care guidelines in CDG? Results from the 4th World Conference CDG Survey. By CDG & Allies - PPAIN.
12.35 - 13.05	Think Tank 6	<p>Keynote Moderator: Prof. Rita Barone and Prof. Ágata Fiumara Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Dorinda Marques da Silva, Dulce Quelhas, Pina Andreotti, Ruqaiyah Altassan, Carlota Pascoal.</p> <ul style="list-style-type: none"> • What are the 4 main obstacles to CDG diagnosis? (10’). • What are the 4 main priorities for CDG care and outcomes? (10’). <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 4 main obstacles and priorities to address CDG diagnosis and clinical impact and devise a short term plan of action. (20’). 	
13.05 - 14.10	Lunch		

WORKSHOP 7: CDG therapies & clinical trials

14.10 - 14.20	Therapies & Clinical trials	Advances of Orpha Labs for Glycosylation Diseases. By Alp Basat Orpha Labs, USA.
14.20 - 14.30		Liposomes for CDG Patrice Rioux Glycomine, USA



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Hours	Session Topic	Theme	Title/ Speaker
14.30 - 14.40	Therapies & Clinical trials		Chaperones for PMM2-CDG. By Belén Pérez CBM-SO, ES.
14.40 - 14.55			Dietary therapies. By Petter Witters UZ Leuven, BE.
14.55 - 15.10			Galactose clinical trial. By Eva Morava Mayo Clinic, USA.
15.10 - 15.25			Is drug repositioning the future? By Mercedes Serrano Hospital Sant Joan de Déu, ES.
15.25 - 15.35			Key lessons for PMM2-CDG PeriQuest. By Ethan Perlstein Perlara company, USA.
15.35 - 15.45			Where do we stand for therapies for CDG? Results from the 4th World Conference CDG Survey. By CDG & Allies - PPAIN.
15.45 - 16.25	Think Tank 7	<p>Keynote Moderator: Prof. David Cassiman and CDG & Allies PPAIN researcher (TBC) Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Alp Basat, Mercedes Serrano, Belén Pérez, Patrice Rioux.</p> <ul style="list-style-type: none"> • What are the 4 main obstacles to therapies and clinical trials for CDG? (10') • What are the 4 main priorities to therapies and clinical trials for CDG? (10'). <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 3 main obstacles and priorities to address regarding therapies and clinical trials in the field of CDG and devise a short term plan of action. (20'). 	
16.25 - 16.55	Coffee break and Poster viewing and presentation		

WORKSHOP 8:

Tackling rare diseases challenges with international and interdisciplinary networks

16.55 - 17.10	International networks		MetabERN: A European reference network to facilitate access to the best care and to boost therapies for CDG. By Maurizio Scarpa MetabERN, IT.
17.10 - 17.20			Share4rare project (remote presence). By Begoña Nafría Hospital Sant Joan de Déu, ES.
17.20 - 17.30			Where do we stand international networks for CDG? Results from the 4th World Conference CDG Survey and major conclusions from Satellite meeting held with professionals on 25 July 2019. By CDG & Allies - PPAIN.
17.30 - 17.40			Title to be defined. By David Coman The Wesley Hospital Brisbane Queensland, AU.
17.40 - 18.30	Think Tank 8	<p>Keynote Moderator: Prof. Luísa Barros Audience will be split up in several working groups that will work on the following questions based on the previous background given.</p> <p>Moderators per working group: Rita Francisco, David Coman, Maurizio Scarpa.</p> <ul style="list-style-type: none"> • What are the 4 main advantages of CDG international networks? (10'). • What are the 4 main priorities of CDG international networks? (10'). <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> <p>Keynote Moderator:</p> <ul style="list-style-type: none"> • Will help to reach consensus as for the 3 main advantages and priorities of international networks in the field of CDG and devise a short term plan of action. (20'). 	
18.30 - 19:00	Break (time to talk and network)		
19.00 - 19.40	Closing session Announce poster winners Surprises from the organizers		
19.40 - 22.00	Dinner & Social Event		

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The Portuguese Association for CDG
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