

Preliminary Agenda

25th July 2019

| Hours | Session Topic | Theme | Title/ Speaker |
|---------------|------------------|----------------|--|
| 14.00 – 14.30 | CDG Nomenclature | What is a CDG? | State of the art for CDG Nomenclature Carlos Ferreira NIH, USA |
| 14.30 – 18.30 | | | <u>Discussion session</u> The Need for Unification: Challenges of CDG Nomenclature |

26th July 2019

| Hours | Session Topic | Theme | Title/Speaker |
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| 8.45 – 9.30 | Registration | | |
| 9.30 – 9.45 | Welcome session Vanessa Ferreira (Organizing committee) | | |
| WORKSHOP 1: Research and development | | | |
| 9.45 – 9.55 | Pharmaceutical drug development in Rare diseases | | Pharmaceutical drug development in Rare diseases at a glance Rita Francisco UCIBIO, CDG & Allies -PPAIN, PT |

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| 9.55– 10.10 | Biobanks and Biomarkers in Rare Diseases | How Biobanking helps the Rare Disease community Speaker to be defined Affiliation to be defined |
| 10.10 – 10.25 | | Biomarkers as invaluable tool for CDG diagnosis and therapeutic advances Speaker to be defined Affiliation to be defined |
| 10.25 -10.35 | | Where do we stand as for CDG Biobanks and Biomarkers at International level? Speaker to be defined Affiliation to be defined |
| 10.35 – 11.15 | Think Tank 1 | <p>Keynote Moderator: Jaak Jaeken</p> <p>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. (10') <p>Short term plan of action to address CDG Biobanks and Biomarkers. (10')</p> |
| 11.15-11.45 | Coffee break | |

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

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| 11.45 – 12.00 | Patient Registries | Patient Registries (PRs) in the field of rare diseases. Speaker to be announced RD-Connect |
| 12.00 – 12.10 | | Case study 1: A European patient registry for rare metabolic disease Maurizio Scarpa MetabERN |
| 12.10 – 12.20 | | Case study 2: CDG Connect Andrea Miller CDG CARE, USA |
| 12.20 – 12.35 | Natural History Studies (NHS) | Case study 1: CDG Natural History Study sponsored by Glycomine Eva Morava Mayo Clinic, USA |
| 12.35 – 12.45 | | Case study 2: NIH CDG Natural History study Lynne Wolfe NIH, USA |
| 12:45 – 12:55 | | Where do we stand as for CDG PRs and NHS at International level? Speaker to be announced |
| 12.55 – 13.35 | Think Tank 2 | Keynote Moderator: Marc Patterson Mayo Clinic, USA Audience will be split up in several working groups that will work on the following questions based on the previous background given. Moderators per working group: Agnes Rafalko, Tomas Honzik, Lynne Wolfe, Peter Witters, Eva Morava <ul style="list-style-type: none"> • What are the 4 main obstacles for CDG Patient Registries and Natural History |

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| | | <p>Studies (NHS)? (10')</p> <ul style="list-style-type: none"> • What are the 4 main solutions for 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). (10') • Short term plan of action to address CDG Patient Registries and Natural History Studies (NHS). (10') |
| 13.35 – 14.40 | <i>Lunch</i> | |
| WORKSHOP 3: Emerging tools/methods to accelerate therapeutic discovery | | |
| 14.40 – 14.50 | Systems Biology for therapeutic discovery | <p>How Systems Biology can boost therapies for CDG and related diseases?</p> <p>Teresa Sardon Anaxomics, ES</p> |
| 14.50 – 15.05 | Artificial Intelligence (AI) | <p>Artificial Intelligence (AI): an introduction</p> <p>Gonçalo Valadao Autónoma University Lisbon, PT</p> |
| 15.05 – 15.15 | | <p>Artificial Intelligence (AI) to accelerate orphan drug development</p> <p>Speaker to be announced</p> |

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| 15.15 – 15.25 | | <p>Case study 1:</p> <p>Artificial intelligence at the service of CDG diagnosis: Early predictive dysmorphic features of PMM2-CDG</p> <p>Antonio Martinez-Monseny Hospital Sant Joan de Déu, ES</p> |
| 15.25 – 15.40 | Metabolomics | <p>How can Metabolomics accelerate CDG and related diseases therapies?</p> <p>Antonio Pineda Hospital Universitari i Politècnic la Fe, ES</p> |
| 15.40 – 16-20 | Think Tank 3 | <p>Keynote Moderator: to be announced</p> <p>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"> • How could these emerging tools/methods to accelerate therapeutic discovery What are the 4 main solutions/areas of impact that these tools can promote (10') • What are the 4 main challenges/obstacles for further implementing and developing these tools in biomedicine? (10') <p>Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience.</p> |
| 16.20 – 16.45 | Coffee break | |
| WORKSHOP 4: CDG models | | |
| 16.45 – 17.00 | <i>In vitro</i> models | <p>European Bank for induced pluripotent Stem Cells (EBiSC) or Modelling diseases process in dish: Induced pluripotent stem cells (iPSCs) technology</p> |

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| | | Speaker to be announced |
| 17.00 – 17.10 | | Induced pluripotent stem cells (iPSCs) for CDG Speaker to be announced |
| 17.10 – 17.20 | | The yeast and CRISPR/Cas to help therapeutic discovery on CDG François Foulquier Université des Sciences et Technologies, FR |
| 17.20 – 17.40 | In vivo model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research | Zebrafish model for PMM2-CDG Paola de Haas Radboudumc, NL |
| 17.40 – 17.50 | | Mouse model for PMM2-CDG Andrew Edmondson Children’s Hospital of Philadelphia, USA |
| 17.50 – 18:00 | | Where do we stand as for CDG disease models at International level? Speaker to be defined |
| 18.00 – 18.40 | Think Tank 4 | Keynote Moderator: to be announced Audience will be split up in several working groups that will work on the following questions based on the previous background given. Moderators per working group: Ethan Perlstein, François Foulquier, Alessandra Cambi, Andrew Edmondson <ul style="list-style-type: none"> • What are the 4 main obstacles for in vitro and in vivo model organisms in the field of CDG ? (10') • What are the 4 main priorities for in vitro and in vivo model organisms in the field of CDG ? (10') Each Moderator per group will share the main conclusions to the Keynote moderator and overall audience. Keynote Moderator: |

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| | | <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. (10') • Short term plan of action to address CDG models. (10') |
| 18.40 – 19.00 | Break (time to talk and network) | |
| 19.00 – 19.40 | Surprise Talk | |
| 19.40 – 22.00 | Dinner & Social Event | |

27th July

| Hours | Session Topic | Theme | Title/Speaker |
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| 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? Alastair Kent UK |
| 9.20 – 9.30 | | | How did Ceracor do it? The case study of FDA approved CDG therapies Perry Calias Cerecor Inc., USA |
| 9.30 – 9.40 | | | How the patient community can be empowered to get involved in drug development Inês Alves PT |
| 9.40 – 10.20 | Think Thank 5 | Keynote Moderator: to be announced Audience will be split up in several working groups that will work on the following questions | |

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| | | <p>based on the previous background given.</p> <p>Moderators per working group: Alastair Kent, Perry Calias, Inês Alves</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. (10') • Short term plan of action. (10') |
| 10.20 – 10.50 | Coffee break | |
| WORKSHOP 6: CDG diagnosis & clinical impact | | |
| 10.50 – 11.00 | CDG Diagnosis | <p>Challenges in CDG diagnosis</p> <p>Dorinda Marques-da-Silva APCDG, PT</p> |
| 11.00 – 11.10 | | <p>Opportunities for CDG diagnostics</p> <p>Dulce Quelhas Centro Hospitalar Universitário do Porto</p> |
| 11.10 – 11.20 | Clinical impact and outcomes | <p>Genetic modifiers for PMM2-CDG diagnostics</p> <p>Giuseppina Andreotti Istituto di Chimica Biomolecolare, IT</p> |
| 11.20 – 11.30 | | <p>Movement disorders as clinical features of CDG</p> <p>Rita Barone UNICT, IT</p> |
| 11.30 – 11.40 | | <p>ImmunoCDGQ results</p> <p>Rita Francisco UCIBIO, CDG & Allies -PPAIN, PT</p> |
| 11.40 – 11.55 | | <p>PMM2-CDG guidelines and standards of care</p> |

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| | | Ruqaiyah Altassan McGill University Health Centre, CA |
| 11.55 – 12.05 | | Outcome assessment tools for rare diseases Carlota Pascoal CDG & Allies PPAIN, PT |
| 12.05 – 12.15 | | How CDG impacts patients and family members. Sum up of coping strategies in CDG families Luísa Barros University of Lisbon, PT |
| 12.15 – 12.25 | | What do we know about and where do we stand for QoL and clinical care guidelines in CDG? Speaker to be defined |
| 12.25 – 13.05 | Think Tank 6 | <p>Keynote Moderator: Rita Barone 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 Barone (keynote moderator), 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 for CDG diagnosis? (10') • What are the 4 main priorities in terms of clinical impact, care and outcomes 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 4 main obstacles and priorities to address CDG diagnosis and clinical impact. (10') • Short term plan of action to address CDG diagnosis and clinical impact/care/outcomes (10') |
| 13.05 – 14.10 | Lunch | |

WORKSHOP 7: CDG therapies & clinical trials

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| 14.10 – 14.20 | Therapies & Clinical trials | Advances of Orpha Labs for Glycosylation Diseases Alp Basat Orpha Labs, USA |
| 14.20 – 14.30 | | Liposomes for CDG Speaker to be appointed Glycomine, USA |
| 14.30 – 14.40 | | Chaperones for PMM2-CDG Belén Pérez CBM-SO, ES |
| 14.40 – 14.55 | | Dietary therapies Petter Witters UZ Leuven, BE |
| 14.55 – 15.10 | | Galactose clinical trial Eva Morava Mayo Clinic, USA |
| 15.10 – 15.25 | | Is drug repositioning the future? Mercedes Serrano Hospital Sant Joan de Déu, ES |
| 15.25 – 15.35 | | PMM2-CDG PerlQuest Ethan Perlstein Perlara company, USA |
| 15.35 – 15.45 | | Where do we stand for therapies for CDG? Speaker to be announced |
| 15.45 – 16.25 | Think Tank 7 | <p>Keynote Moderator: to be announced</p> <p>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</p> <ul style="list-style-type: none"> • What are the 4 main obstacles for therapies and clinical trials in the field of CDG ? (10') |

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| | | <ul style="list-style-type: none"> What are the 4 main priorities for therapies and clinical trials 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 therapies and clinical trials in the field of CDG. (10') <p>Short term plan of action. (10')</p> |
| 16.25 – 16.55 | Coffee break | |
| WORKSHOP 8: Tackling rare diseases challenges with international and interdisciplinary networks | | |
| 16.55 – 17.05 | International networks | <p>CDG & Allies PPAIN: An International patient founded network to accelerate translational therapeutic research.</p> <p>Rita Francisco CDG & Allies PPAIN, PT</p> |
| 17.05 – 17.20 | | <p>MetabERN: A European reference network to facilitate access to the best care and to boost therapies for CDG</p> <p>Maurizio Scarpa MetabERN, GE</p> |
| 17.20 – 17.30 | | <p>Share4rare project</p> <p>Begoña Nafría Hospital Sant Joan de Déu, ES</p> |
| 17.30 – 17.40 | Communication & Awareness | <p>Speaking the same language: The role of social media for Rare Disease awareness</p> <p>Eleonora Passeri IT</p> |
| 17.40 – 18.20 | Think Tank 8 | <p>Keynote Moderator: to be announced</p> <p>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, Maurizio Scarpa, Eleonora Passeri</p> <ul style="list-style-type: none"> What are the 4 main advantages of international networks in the field of CDG ? (10') |

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| | <ul style="list-style-type: none"> • What are the 4 main priorities for international networks 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 advantages and priorities of international networks in the field of CDG. (10') • Short term plan of action. (10') |
| 18.20 – 18:35 | <i>Surprise Talk</i> |
| 18.35 – 18.50 | Closing session/Final conclusions Vanessa Ferreira (+ Organizing committee) |
| 18.50 – 19.20 | <i>Break</i> (time to talk, relax and network) |
| 19.20 – 22.00 | <i>Dinner & Social Event</i> |