FAMILIES AND PROFESSIONALS UNITED TO TACKLE THE THERAPEUTIC NEEDS OF CONGENITAL DISORDERS OF GLYCOSYLATION

Authors: Pascoal C\textsuperscript{abc}, Francisco R\textsuperscript{abc}, Brasil S\textsuperscript{abc}, Marques-da-Silva D\textsuperscript{abc}, Videira P\textsuperscript{abc}, dos Reis Ferreira V\textsuperscript{abc}.

Affiliations: (a) UCIBIO, Departamento Ciências da Vida, Faculdade de Ciências e Tecnologia, Universidade NOVA de Lisboa, Portugal, (b) Portuguese Association for CDG, Lisbon, Portugal; (c) CDG & Allies – Professionals and Patient Associations International Network (CDG & Allies – PPAIN), Portugal

INTRODUCTION

Congenital Disorders of Glycosylation (CDG) are one of the fastest growing groups among the 8000 rare diseases currently known. There are more than 140 reported CDG so far which are caused by mutations in glycosylation-related genes. CDG patients often present multi-systemic phenotypes and high clinical inter and intra heterogeneity.\textsuperscript{1}

To overcome the challenges of CDG research (Fig.1), a unique, patient-led, multistakeholder infrastructure to foster investigation and better care in CDG as been created – the CDG & Allies – Professionals and Patient Association International Network (CDG & Allies – PPAIN).

AIM

Here, we describe how CDG & Allies – PPAIN is fomenting patient-centric research. The exceptional case study of the Glycoimmunology Working Group (WG) is highlighted.

CDG & ALLIES – PPAIN

CASE STUDY: WG CDG & GLYCOIMMUNOLOGY

- FACT: 20% of child mortality in PMM2-CDG (most frequent type) is due to recurrent and severe infection\textsuperscript{b}.

- PATIENTS’ NEED: Targeted and effective therapy.

1. Co-create and administer a patient/caregiver-directed questionnaire to access the immunological issues in CDG.

2. Accelerate the access to patient samples and perform wet lab research to identify underlying ocular/cellular mechanisms and therapeutic targets.

3. Gather funds to enable research on the cellular and molecular mechanisms to find potential therapeutic targets.

CONCLUSION

CDG & Allies–PPAIN is showing that working for and with patients is key to provide better, faster and more focused avenues to therapeutic development.

REFERENCES


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For further information, please contact the Portuguese Association for CDG (APCDG), CDG&Allies-PPAIN and World CDG Organization.

Email: sindromecdg@gmail.com | www.apcdg.com | www.researchcdg.com | www.worldcdg.org

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