ICPGC April 2022 Newsletter

Hello ICPGC members,

Since our last issue we have had two virtual meetings. The October meeting was hosted by the McLaughlin Centre and supported by CanChild and CP-Net, and our recent March meeting was hosted by the Australasian Academy of Cerebral Palsy and Developmental Medicine. Both meetings had a great selection of talks that looked at genome findings from large research cohorts, perspectives from families, experiences in clinical genetics, extensive functional validation research, and the role of epigenetics in CP. For those who registered to attend the AusACPDM workshop, video recordings are available until May 5th. The Governance Council are currently planning for a 2023 meeting and will pass on details as soon as possible.

Finally, Charles Steward (Congenica) from the Governance Council recently published a piece with Epilepsy Research UK about his journey in the field of genomics and how genetic data can be transformative in diagnosing, treating, and preventing health conditions. Find the blog post here.

ICPGC Governance Council

P.S You can find all previous copies of our newsletters online here at ICPGC.org
P.S If you have friends / family members / colleagues who would like to join the ICPGC, they can register here. Or subscribe to this newsletter here.

RECENT RESEARCH HIGHLIGHTS

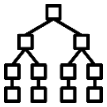
Yana Wilson (CPA, University of Sydney) and the **ICPGC Phenotype Working Group** have published their work on developing Common Data Elements for genomics studies in cerebral palsy. To read more, click here. For the most up to date version of the CDEs, please see here.

Siddhartha Srivastava (Boston Children's Hospital) and colleagues performed comprehensive phenotyping and WES on a cohort of 50 individuals with cryptogenic CP, non-cryptogenic CP, and CP masqueraders. Twenty-six percent of the cohort had a pathogenic/likely pathogenic variant in 13 unique genes. To read more, please click here.

Michael Kruer (Boston Children's Hospital, University of Arizona), **Saima Riazuddin** (University of Maryland) and colleagues report on 28 bi-allelic variants in SPATA5L1 associated with sensorineural hearing loss with or without a nonprogressive, complex neurodevelopmental phenotype. To read more, please click here.

Jan Friedman (University of British Columbia), **Peter van Essen** (Radboud University), and **Clara van Karnebeek** (University of British Columbia, Radboud University, Amsterdam University) have produced a comprehensive review of the genetic literature of CP. To read more, please click here.

Bhooma Aravamuthan (Washington University) and colleagues surveyed 197 members of the CP community on their views of having a CP diagnosis, an etiological diagnosis (specifically genetic), or both. To read more, please click here.



Gareth Baynam (Western Australian Register of Developmental Anomalies) and colleagues have had a manuscript accepted into Nature Biotechnology on standard for phenotypic exchange. The Phenopacket schema is a freely available, community driven standard that

streamlines exchange and systematic use of phenotypic data and will facilitate sophisticated computational analysis of both clinical and genomic information to help improve our understanding of diseases and our ability to manage them. Find the preprint here.

ICPGC GENERAL UPDATES

NGLY1 Deficiency

During the ICPGC Virtual Summit in October 2021, Ms Caroline Stancliff from Grace Science LLP spoke about key phenotypes to look out for in people who may have an NGLY1 Deficiency. These include alacrima or hypoalacrima, global developmental delay, movement disorder, and transient elevation of ALT & AST. Grace Science LLP is seeking to identify more patients with this genetic variant, please see the Clinician Handout for more information, including their contact details.

CP Commons is now live

The CP Commons was established to centralise information and resources necessary to drive the discovery, interpretation, and characterisation of the genome's role in cerebral palsy. To accompany the launch of the CP Commons, we have developed a Launch Report, which offers both an overview of the platform as well as a user guide for researchers interested in becoming members. All members are welcome to register for an account here.

ClinGen Gene Curation Expert Panel – Cerebral Palsy

The ClinGen framework gives an evidence-based approach to gene-disease curation via a specific schema for objectively scoring genetic and experimental data. The Cerebral Palsy CGEP has been approved by ClinGen and will commence curation of genes identified in people with CP. If you are interested in joining this group, please contact Andres Moreno De Luca (amorenodeluca@geisinger.edu).

Australian Panel App – Cerebral Palsy

The Australian PanelApp is a crowdsourcing tool to allow gene panels for specific conditions to be shared, downloaded, viewed, and evaluated by the scientific community. Clare van Eyk (Adelaide University) and Zornitza Stark (Australian Genomics) have established a gene panel dedicated to CP, which can be found here. For more information on how you can contribute to this resource, please click here.

Human Phenotype Ontology – Cerebral Palsy

Gareth Baynam & Dylan Gration (Western Australian Register of Developmental Anomalies) are collaborating with Peter Robinson, at the Jax Institute, and other colleagues on enhancing Cerebral Palsy HPO terms.