ICPGC October 2021 Newsletter

Hello everyone,

This week we celebrate World CP Day on October 6th!! World CP Day gives us the opportunity to come together to celebrate and support those living with CP, embrace diversity, and to help create more awareness and action around CP.

In recognition of World CP Day, this newsletter includes information about registering for the 4th ICPGC Annual Meeting, the 8th Annual CP-NET Science and Family Day, the AACPDM PTC Therapeutics Symposium, as well as some recent genomics research highlights from members of the consortium and beyond.

We look forward to seeing you all at the 2021 ICPGC Virtual Meeting in a couple of weeks.

ICPGC Governance Council

P.S If you have friends/family members/ colleagues who would like to receive this Newsletter please let them know that they can subscribe here.

MEETINGS

2021 4th ICPGC Meeting – October 22 and 23

The 2021 Annual meeting is a little more than two weeks away. Details:

- Friday October 22 and Saturday October 23 at 6:00am-9:00am Toronto (EST).
- On Zoom
- Registration is free and now OPEN. To register, please click here. Please register by October 16.
- A meeting program will be made available via the ICPGC.org soon.

The keynote lecture will be given by Prof David Rowitch, Professor of Paediatrics and Head of Department, University of Cambridge (UK). Professor Rowitch will give his perspective as both a neonatologist and neuroscientist about human neurobiology that pertains to CP genetics and transcriptomics, the clinical application of genome sequencing in the UK's National Health Service, and insights into genetic conditions that are misdiagnosed as CP early in life.

2021 8th Annual CP-NET Science and Family Day – October 23

CP-NET is excited to invite families, caregivers, people with CP, healthcare professional and researchers to join us for a day of research and celebration in recognition of World CP Day. This meeting is open to all, and all presentations will be given in plain language with a family audience in mind. Details:

- Saturday October 23 at 10:00-3:00pm Toronto (EST).
- On Zoom
- Registration is free and now OPEN. To register, please click here. Register by October 16.

The keynote lecture will be given by Dr Evelyn Constantin, Associate Professor from McGill University. Dr Constantin's research focuses on sleep in children, the impact of treatment for sleep disorders on specific health outcomes, and the promotion of healthy sleep habits.

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2021 AACPDM PTC Therapeutics and Invitae present Real-World Experience with a No-Charge CP Genetic Testing Program – October 12

AACPDM are offering virtual product theatres this year. Tim Feyma (Gillette Children's Specialty Hospital), Warren Marks (Cook Children's Medical Centre) and Michelle Fox (Invitae) discuss their real-world experience with a no-charge cerebral palsy genetic testing program. The symposium will cover the provide an overview of the PTC Pintpoint™ Cerebral Palsy Spectrum program, explore case studies of patients who had an underlying genetic etiology, and present initial real-world data from the program. This symposium is restricted to licensed healthcare professional only.

- Tuesday October 12 at 7:00pm EST
- Virtual
- Registration is now OPEN. To register, please click here.

2022 AusACPDM/IAACD/5th ICPGC Virtual Meeting

Due to the ongoing challenges of the COVID-19 outbreak in Australia, the AusACPDM Board and the IAACD Steering Committee have made the difficult decision to make the AusACPDM/IAACD fully virtual in March 2022. We are now working with the committee to finalise times for the 5th ICPGC meeting. We will distribute more information in due course.

RECENT RESEARCH HIGHLIGHTS

Yana Wilson (Cerebral Palsy Alliance, the University of Sydney) and colleagues have published a study about what people with CP and their families think about genomics research. The study found that willingness to participate in genomics research was associated with tertiary education, previous genetic testing experience, overall higher genomics awareness, and trust in international researchers. Furthermore, while participants are supportive of secondary data-use or biobanking activities, people with CP and their families still wanted to be informed about these activities, even if the decision to access their information is granted by a third party. Continued consultation and engagement with individuals with CP and their families is imperative to promoting trust, awareness, and ongoing participation and support.

To access the article, please click here: https://www.karger.com/Article/PDF/518942

Clare van Eyk (University of Adelaide) and the group from Adelaide recently published a study that assessed the yield of clinically reportable genetic variants identified using WGS in an unselected cohort of people with CP. The study, published in Genomic Medicine, detected pathogenic or likely pathogenic variants in nearly a quarter of the participants (24.7%, n=37). A further 34.7% carried variants of unknown significance in variation intolerant genes. Of note, the authors highlighted that approximately 50% of the participants would likely benefit from a change in clinical management because of the genetic findings.

To access the article, please click here: https://www.nature.com/articles/s41525-021-00238-0

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Qinghe Xing (Fudan University), **Changlian Zhu** (Zhengzhou University) and colleagues have published a study in Journal of Genetics and Genomics, where they have screened a cohort of people with CP (n=667) and controls (n=573) for variants in a possible novel CP gene, Telomerase associated Protein 1 (TEP1).

To access the article, please click here:
https://www.sciencedirect.com/science/article/pii/S1673852721002824?via%3Dihub

Somayeh Bakhtiari (Phoenix Children's Hospital) and colleagues published a study in Genomics Medicine after identifying six patients from three unrelated families with variants in a novel CP gene, Nuclear Speckle Splicing Regulator Protein (NSRP1). Phenotypic features present include developmental delay, epilepsy, microcephaly, hypotonia, and spastic cerebral palsy.

To access the article, please click here: https://pubmed.ncbi.nlm.nih.gov/34385670/

An excellent paper was recently published in Molecular Genetics and Metabolism by A. Elliot & C. Guimond that reviews recent advances in sequencing for CP, describes clinical features suggestive of genetic etiology of CP, practice guidelines for genome-wide sequencing of individuals with CP, and a practical approach to the genetic counselling of these families.

To access the article, please click here:
https://www.sciencedirect.com/science/article/pii/S1096719221007551?via%3Dihub

If you have any updates or research news that you want to share, please send these through to info@icpgc.org, so they can be included in the next newsletter.