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Engaging families in research to drive progress: Phelan-McDermid Syndrome International Registry (PMSIR) and the Phelan-McDermid Foundation Data Network (PMS_DN)

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Phelan-McDermid Syndrome (PMS) is a rare genetic condition associated with autism spectrum disorder, seizure disorders and severe to profound intellectual disability. Today, there is no cure for PMS and patient interaction with health care and research is for the most part navigated by parents and caregivers. In 2011, the PMS Foundation launched the PMS International Registry (PMSIR), centralizing data about the PMS community and removing barriers for researchers studying the condition and its associated interventions. The PMSIR has been family-led since its inception. In 2013, the Foundation was awarded a PCORI contract to participate in PCORnet as a Patient Powered Research Network and establish the PMS Data Network (PMS_DN), integrating patient-reported outcomes from the PMSIR with concepts extracted from electronic health records of PMS patients. The PMS_DN, a collaboration between the PMS Foundation, Harvard Medical School Center for Biomedical Informatics and Boston Children's Hospital, advances knowledge, care and treatment of PMS and related conditions by integrating diverse, complex data sources into a richly populated, high quality and centralized database to facilitate patient-centered research. The PMS_DN technical infrastructure is an i2b2/tranSMART data warehouse and web interface, which integrates patient reported outcomes (PROs), curated genetic testing results and knowledge extracted from clinical notes. The PMS_DN excels in engaging families in data sharing activities and prioritizing research questions, facilitating family communication and promoting transparency of patient data use in research, through the leadership of exceptional parents and the authentic engagement of patients and caregivers as champions for their families.

Biography

Megan O'Boyle is the Principal Investigator for the Phelan-McDermid Syndrome Data Network (PMS_DN, PCORnet) and the Phelan-McDermid Syndrome International Registry (PMSIR). She is passionate about the value of the patient's voice in research, drug development, clinical trial design, development of related legislation and quality of life decisions. She advocates for data sharing, collaborating with other advocacy groups, sharing resources and streamlining IRB practices and policies.

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Notes:

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Page 31