Background about the Phelan-McPosium

The Phelan-McPosium: A Patient Centered Outcomes Workshop occurred on Thursday, July 21 and Friday, July 22, 2016 in Orlando, FL as part of the Phelan-McDermid Syndrome Foundation (PMSF) 2016 International Family Conference. The mission of PMSF to improve the quality of life of people affected by PMS by providing family support, accelerating research, and raising awareness. The primary goal of this conference and the inaugural Phelan-McPosium is to embrace all attendees—individuals with PMS, family members and caretakers of individuals with PMS, researchers, clinicians—as part of the PMS community and work collaboratively to identify the areas where growth is needed in PMS research and practice.

PMS is a rare genetic condition caused by deletions of 22q13 or mutations of the SHANK3 gene resulting in a highly heterogeneous phenotypic presentation often including developmental disability, autism, hypotonia, and other complex medical and psychiatric conditions. Because the condition is very rare (only 1300 known cases worldwide), the majority of doctors treating these patients have limited awareness and understanding of the genetic syndrome and its complex presentation. Thus, parents and/or other caretakers have a vital role in educating medical professionals and researchers about PMS. Unfortunately, parents often do not have access to the published literature about PMS and/or do not have the background training to translate this information into practice. Furthermore, researchers studying PMS have limited access and awareness of the families and their needs. Thus, the need for increased collaboration and communication between families, providers, and researchers is paramount for the health and well-being of individuals with PMS and their families.

Thus, the questions that guided the establishment of the Phelan-McPosium were: 1) how can family members communicate their concerns and priorities to the medical and scientific community and have a role in the design and conduct of future research studies and 2) how can family members receive information to help them make the appropriate medical, behavioral, and educational decisions together with their children's medical professionals?

The Phelan-McPosium aimed at providing up-to-date research to families and caretakers as well as facilitating discussions between the families and researchers/clinicians in order to identify research gaps and priorities. The primary goal of the Phelan-McPosium was to give caregivers the opportunity to engage with researchers and other families so that they felt empowered to handle the challenges of caring for someone with PMS. This goal is directly in line with the 10th biennial conference, "Embrace, Engage, Empower". Furthermore, because this is the largest gathering of families affected by Phelan-McDermid Syndrome (PMS) and researchers studying PMS worldwide, participants are afforded the opportunity to 1) network with

families, researchers, clinicians, 2) participate in social activities and support, 3) learn from educational opportunities with leading experts within the field, and 4) discuss their concerns in an open and safe environment.

Within the Phelan-McPosium, seven one-hour sessions centered on specific topics that were selected based upon their high relevance to quality of life (QOL) to individuals with PMS and their families. The topics included: 1) genetics, 2) cognition, learning, and development, 3) communication and social behavior, 4) sleep issues, 5) gastrointestinal issues, 6) lifespan and natural history, and 7) epilepsy. Each topic included a 20-minute presentation by one of the leading experts within the field, a 20-minute parent-led roundtable discussion between family members and researchers/clinicians, and concluded with a 20-minute panel discussion by experts working within each of these areas to address any further questions as well as discuss potential future projects.

The introductory presentation provided family members lay-friendly scientific information about what has already been learned in the topic with the aim of helping families make appropriate decisions regarding medical, behavioral, and educational issues affecting individuals with PMS. During the roundtable portion, a parent facilitator led discussions regarding their experiences within the topic area and posed questions to other parents and researchers/clinicians. The parent facilitator recorded pertinent details from the discussion and questions. During the roundtable portion, family members also participated in online polling through Poll Everywhere. The polls also were available to families who are not able to attend the meeting. Finally, during the panel of experts, the ideas and priorities of the families, discussions about current projects, and possible future study ideas were discussed.

Many of the experts and panelists were researchers in two new PMS clinical research consortia, and their firsthand participation in the workshop was informative to the development of new research questions and studies. While this is not the first time PMSF brought researchers and families together, it was the first structured opportunity for parents to be partners in the design and conduct of new research studies. Overall, the format of the Phelan-McPosium was designed for families to be able to discuss with researchers/clinicians real and pertinent issues faced by individuals with PMS and their caretakers, with the ultimate goal of informing the development of new research.

The short-term aims of the Phelan-McPosium were two-fold: 1) To collect first-hand information from families regarding their experience, questions, and priorities and, 2) to return lay-friendly, current research findings back to families.

The long-term aims of the Phelan-McPosium were two-fold: 1) to shape the development of new research studies which reflect the greatest concerns and priorities of families and lead to the development of new treatments, therapies, and/or clinical guidelines for individuals with PMS, and 2) to help families make the best decision for their children.

The white papers are structured in the following ways. First, a white paper was written separately for each pre-identified topic (e.g., genetics, epilepsy). Each whitepaper begins with a summary of the information provided during the expert presentation. Next, three to four issues within each topic were identified as primary concerns by the parents. Then, the white paper concluded with proposed solutions for each of these issues.

Several factors were considered in identifying the primary issues raised by parents. First, we used the Poll Everywhere (PE) questions as an informal guide. The PE ratings were used as metrics of importance of the topics being discussed. For example, if a particular question only garnered 10% concern or prevalence, it may be omitted. On the other hand, if a question indicated 90% concern or prevalence, this may be considered a high priority/issue. Second, because parent facilitators were took detailed notes during the discussion, we used these notes to count "new" or previously unmentioned concerns. Additionally, these notes also could have provided further evidence for or against PE questions. We examined these roundtable findings across tables to determine the concordance among tables. We also compared the facilitators' notes with the polling results to determine if there was concordance between roundtable findings and PE findings. For each table, a tally of how many parents indicated an item to be a concern was computed as well as the average rating of concern. Level of concern was rated 1 – 5, with 1 being a low concern and 5 being a very significant concern. Although this allowed us to quantify concerns, this metric is imperfect as not all facilitators consistently took counts of parents' concerns or asked about severity rating. Thus, the metrics we provide in the white papers should be interpreted with some caution, as they are estimates. However, because not all tables counted and rated parent concerns, we these estimates are likely an under-estimate of true concern. Third, any new or repeated questions raised during the panel discussions also were considered. Finally, based upon the prevalence and severity of concern according to PE polls and roundtable and panel discussion, we were able to identify three or four primary issues within each topic. The white papers were arranged in such a manner that the concern of highest degree was presented first, and the second highest second, and so forth.

The proposed solutions were developed based upon: 1) information from experts during the presentation and panel discussion, 2) discussion during the Consensus meeting that followed the McPosium consisting of lead researchers and clinicians, 3) ideas and practices used in other neurodevelopmental disorders and genetic syndromes. It should be noted that the proposed solutions are by no means exhaustive, but importantly highlight potential ideas for the future and should serve as a guideline for next steps.