

Important Message from our Medical Advisory Committee

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From: Alex Kolevzon, MD, PMS MAC chairperson; William Bennett, Jr, MD, MS; Elizabeth Berry-Kravis, MD, PhD; Ann Neumeyer, MD; Curtis Rogers, MD; Teresa Kohlenberg, MD

Re: Information for Caregivers of Patients with Phelan-McDermid Syndrome (PMS)

Dear families affected by Phelan-McDermid syndrome,

The Phelan-McDermid syndrome community has experienced profound grief and concern over the deaths of some of our beloved children and adults. The PMS Foundation (PMSF) is urgently working with the PMSF Medical Advisory Committee (MAC) to understand the causes of these deaths and has begun a “mortality review.” This review is a comprehensive and detailed process of speaking with families, reviewing medical records in depth, and identifying causes of death and any patterns that emerge which will be helpful to reduce risk.

In the past five years, PMSF is aware of 35 individuals who have passed away. Their ages have ranged widely from younger than 1-years-old (including at birth) to 52-years-old. Males and females were equally at risk. There is still much to learn, but based on the current state of knowledge, we can say that many deaths have been related to:

- o complications of severe epilepsy
- o choking / aspiration, possibly related to catatonia - associated feeding problems
- o significant worsening of problems with digestive tract functioning
- o presence of a co-existing genetic condition in addition to PMS

Thus, there is likely an increased risk of early mortality in Phelan-McDermid syndrome with its associated health conditions. While this is alarming and there is still much to understand in more depth, the current mortality rate in Phelan-McDermid syndrome known to us is still within reported rates of mortality of infants and adults in the US general population.

PMSF and MAC have planned two steps to better understand this risk.

With the help and consent of the families, we are now collecting all available medical records to corroborate and amplify our understanding. Six families have agreed to participate thus far and we aim to collect records from at least ten. Next, we will conduct a qualitative assessment where we interview families who lost loved ones to gain deeper knowledge of the details and circumstances surrounding the death. Our ultimate goal is to *identify early risk factors* and potential *management strategies* to reduce the risk of mortality. We will also publish and share these findings.

Please contact info@pmsf.org if you have questions or concerns. Details about requesting a consult from the PMS Neuropsychiatric Consultation Group are here:

<https://pmsf.org/neuropsychiatric-consultation-group/>