The indispensable parental perspective on a rare Neurodevelopmental Disorder;

An international survey among 587 PMS families worldwide

Aim:
Integrate the parental perspective on a rare NDD in a European clinical guideline

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Background

Phelan-McDermid syndrome (PMS), a rare Neurodevelopmental Disorder (NDD) caused by deletion or pathogenic variation of the *SHANK*3 gene, has a phenotype of intellectual disability (ID) and concerning psychiatric disorders such as autism, regression and catatonia. The European guideline consortium collected the parental perspective on care needs, somatic and psychiatric issues and parental stress. Integration of the parental perspective and literature reviews led to practical guideline recommendations.

Method

Parents of children with PMS were recruited through patient organizations worldwide on request of the European PMS guideline consortium. The multilingual survey consisted of 35 questions divided into four sections. Based on the survey results, experts on PMS conducted structured literature reviews. The reviews and survey led to recommendations voted on by the consortium based on the AGREE-principles of guideline development.

Survey quotes parents:

"I feel lost. She is becoming a teen and now everything seems to get harder".

"I find that the hardest and scariest part, who will take care of my adult child who will most likely always be a child".

"How to manage all of my child's needs without burning out"?

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Results

A total of 587 surveys from 35 countries was included. A wide variety of developmental, neurological, and other clinical problems was reported. Most frequently experienced problems were related to speech and communication (97%), ID (95%) and behavioral issues (71%).
Developmental regression appeared to begin earlier in this cohort than previously described. Parents reported elevated general parental stress with specific child- and context-related contributing factors. Comparison of the results of this survey with literature on PMS led to new insights on prevalences of mental health and somatic issues.

Conclusion

This work has led to various validated recommendations in the European PMS Guideline including an age specific surveillance scheme, specific genetic counselling, structured evaluations of different health issues and a focus on family well-being.
In developing a guideline on a rare NDD, like PMS, the parental perspective is of the essence.

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