

## **Report from the PMSF Family Conference 2024**

The PMSF organized a family gathering for the first time since 2018. It took place in Bloomington, Minnesota, approximately in the middle of the United States. The conference lasted from July 18-20. More than 800 attended, 150 of them online. Some families from other countries also attended, including Brazil, Canada, France, and Holland. The event was excellently organized and I was warmly welcomed into the community.

The new motto of the PMSF was presented in the form of the three Cs:



### **The current key topics of the PMSF**

All three days had very informative presentations. Dr. Teresa Kohlenberg (neuropsychiatry), Dr. Billy Bennett (digestive disorders), Dr. Chris Winrow (lymphedema; all three are parents of a child with PMS), Dr. Tess Levy (genetics), Dr. Jimmy Holder (epilepsy - by video), gave talks on specific medical problems. Dr. Michael Schön from our family association was invited to present the European guidelines. In the same session, Dr. Sid Srivastava presented the US guidelines. There were also presentations on adult accommodation and dietary recommendations by dietician Chardell Buchanan. Furthermore, a workshop session on augmentative and alternative communication was held.

In the research section, various studies were presented as posters. Dr. Alex Kolevzon was present via video link and provided information about opportunities to participate in clinical studies. Neuren, a pharmaceutical company that conducted a first study with a drug similar to the body's own hormone IGF-1, was the main sponsor of the meeting. They also had some of their staff on site. Jaguar Gene Therapy, a company that has developed a gene therapy for PMS, was also represented. Neuren showed trial results that are promising, as we reported in the newsletter some time ago. They will soon be starting a phase III trial, which could potentially lead to approval if successful. Jaguar and Dr. Guoping Feng have previously carried out animal studies on mice and monkeys, also with positive results. In the fall, they will start their first study on individuals with PMS. This involves injecting a virus, which serves as a gene shuttle, into the inner cavities of the brain, the ventricles. The nerve cells are infected, but no new viruses are formed, only the artificial SHANK3 gene is inserted. Guoping Feng, a renowned neuroscientist who has been researching SHANK3 for a long time, had developed a short variant of the SHANK3 gene that contains essential parts of the gene. This short SHANK3 is used in this therapy. A very interesting presentation was given by Dr. Antonio Persico on a therapy study for PMS with vitamin and coenzyme Q10.

Michael Schön is a member of a planning group for the founding of an international federation of all family organizations (name: Phelan-McDermid Syndrome International Federation, PMSIF). He is currently accompanied by one representative from Spain, one from the United Kingdom and two from the United States. The concept was introduced to representatives from the organizations in France, Italy, Canada, Brazil, and the Netherlands. The idea and objectives of this project were very positively received.

As a social program, there were various activities for the children in the Mall of America, the largest shopping mall in America with its own amusement park and aquarium. We were

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invited by the PMSF to a joint dinner on Thursday evening in the conference rooms. There was also a meeting of the "Founders" of the PMSF with Dr. Katy Phelan and Dr. Curtis Rogers, among others, whom we were able to win as guest experts for the last family meeting in Schönblick. It was very exciting to listen to the story about the early days of the Foundation. There were also very moving contributions from the parents present, who were in tears as they reported how important this community and the support was for them. Very touching was the presentation of a former star fashion photographer, Rick Guidotti, who made it his mission to show the beauty of people with diseases in order to change society's negative attitude towards diseases, especially genetic disorders. He also offered to take pictures of the children and families on location. His project is called Positive Exposure (<https://positiveexposure.org/>). Dr. Kate Still, the scientific director, was bid farewell and thanked for her great contribution to the PMSF.



**Group foto**

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