

## **Largest number of families ever attends**

### **Phelan-McDermid Syndrome conference**

**By Nick Assendelft**

Vice President, Board of Directors

Outstanding. Wonderful. Enriching.

That's how many family members described their experiences at the 2014 Phelan-McDermid Syndrome Foundation International Conference in Orlando, Fla., the largest gathering ever of families affected by Phelan-McDermid Syndrome.

A total of 196 families attended the biennial conference July 23-27 that was focused on helping families from around the world. Families came from 38 U.S. states and 14 countries, including Canada, Germany, the United Kingdom, Costa Rica, Brazil, Mexico and Malaysia.

"What an awesome time we had away," said **Megan Toole** of Australia. "We have made lifelong friendships, gained a better understanding about Phelan-McDermid Syndrome and what this means for our family. Cannot wait for the next conference to do it all over again."

The four-day gathering included discussions of education and lifestyle issues, analysis of the latest research efforts and opportunities to form support networks with others affected by PMS.

"The success of this year's conference was the result of the efforts and participation of many talented and generous people," said **Barbara Cruz**, PMSF Executive Director. "That includes parents, speakers, volunteers, helpers, sponsors and office staff. "Without your help this year's conference would not have been possible."

The conference was a Sweet 16 for the Phelan-McDermid Syndrome support group. It was 1998 when families met for the first time to learn more about the genetic condition caused by a missing or damaged portion of the 22nd chromosome. Families have met every two years since, with the number growing from 23 the first year to 196 in July. The PMS Foundation, based in Venice, Fla., was formed in 2002.

"It was so heartwarming to see all the new families at the conference and reconnect with families," said **Susan Lomas**, President of the Foundation's Board of Directors. "Our Foundation's mission is to support families who have special needs. This conference filled that mission. It was a testament to the strong bonds that so many of us share."

There were sessions for moms (moderated by **Susan Lomas**) and dads (**Ryan Cox** and **David Mancini**), married couples (**Judi** and **Rory Jones**) and single parents (**Cindy Dulaney** and **Kathy Brown**), extended

families (**Debbie Graham**), siblings (**Amber Curell**), new families (**Nick Assendelft, Dr. Katy Phelan and Dr. Curtis Rogers**) and roundtable discussions where families could share advice about raising a child with Phelan-McDermid Syndrome.

Other topics included issues related to aging (**Diane Linnehan and Sally Richards and Judi and Rory Jones**), financial planning (**Rick Wood**), communication (**Patricia Ourand**), legislative advocacy (**Mark Vieth**), family coping (**Evan Farrar** of the Prader-Willi Syndrome Association), organizing medical records (**Megan O'Boyle**), schooling (**Christine Goulbourne**), and yoga and physical therapy (**Tiffany Engle**).

Families could also participate in an art therapy program (run by **Amy Estrela**), corn hole competition (**Melissa and Todd Goldsmith**), free studio photos (**Elizabeth and Trey Homan**), a Sweet 16 dance (**Rachel Assendelft**) and our traditional conference-ending whiffle ball game (**Chris Cruz and Mario Camarillo**). A total of 37 siblings and chaperones bonded on a day trip to Islands of Adventure and participated in Club 22 (**Amber Curell**).

A big part of the conference was devoted to health and science, led by **Geraldine Bliss**. Researchers discussed the latest findings in the progression of PMS, clinical research (**Joe Buxbaum, Alex Kolevzon, Audrey Thurm, Lindsay Oberman, Latha Soorya, Craig Powell, Matt Mosconi and Eva Loth**), an update on biosamples (**Joachim Hallmeyer, David Panchision, Joe Buxbaum, Ruth Ann Luna and Alison Singer**), PMS phenotypes and genotypes (**Alex Kolevzon and Catalina Betancur**), translational research (**Craig Powell**), PMS innovators (**Alex Shcheglovitov, Xiaoming Wang, Sally Kim and Luigi Boccuto**), adult medical issues (**Dr. Katy Phelan**), lymphedema (**Dana Winrow**), seizures (**Jimmy Holder**), toileting (**Judith Coucouvanis**) and autism (**Audrey Thurm**). **Dr. Katy Phelan** and **Catalina Betancur** helped to decipher genetic reports for parents.

Families also learned more details about the Foundation's collaboration with Harvard University on a contract with the Patient Centered Outcomes Research Institute (**Megan O'Boyle, Liz Horn, Andria Cornell, Paul Avillach and Sarita Wahba**). The contract will expand our international patient registry over the next 18 months to better collect medical data about PMS. This will facilitate research projects and guide family doctors treating those with PMS.

The "In It To Win It" raffle, organized by **Terri Dutts** and **Jennifer Mancini**, was once again an overwhelming success, raising more than \$4,500. And we also had 43 items donated for the door prize drawings at the closing session. **Darla** and **Matt Schaefer** won the grand prize of dinner with our founder, **Dr. Katy Phelan**.

Our regional representatives had a chance to meet under the direction of **Teri Dutts**, our membership committee chairperson. The conference also was the setting for the first meeting of our Global Partners. Representatives from 12 countries talked about how best to collaborate on many fronts. **Susan Lomas** has begun to develop this program. PMS groups in other countries are their own associations but are linked together as collaborative partners with the same mission to help our families. Separately, **Catalina Betancur** met with the families from Spain. We also, for the first time, offered translators

during some of our sessions.

A handy crew made sure all the audio-visual connections were working correctly and the live streaming - a hit with families - went off without a hitch. Rich "26.2" Lomas headed the team that also included Eric "E-Tech" Youngquist, Rick "Big Rick" Kuehl, Kenneth "Head Banger" Rosales and Stephanie "Canadian Spice" Leclerc.

One of the more popular features of the conference was our phone app, developed by **Sara Greenlaw-Carlmark**. The app was downloaded 628 times (486 in U.S., followed by 37 in Australia, 30 in the United Kingdom and 19 in Canada). There were roughly 7,639 page views, and the app was used about 15,430 times. Live streaming and agenda pages were used often.

We would also like to thank all the volunteers who over the past two years have helped the Foundation grow, whether through donation of time, money or services. Thank you, too, to our top fund-raisers: **Veronica Frunzi** for her Annie's Golf Classic, **Jennifer Randolph** for her Phelan Lucky nationwide T-shirt sales, **Debbie** and **Mike LeClaire** for Logan's Heroes motorcycle ride and auction, **Megan** and **Michael O'Boyle** for their "\$22 in 22 Days" campaign and **Barbara Cruz** and the PMSF office staff for the Giving Challenge.

The conference also featured our Foundation's new logo, designed by **Troy Scillian**. The logo takes into account all the stakeholders who help make us grow: Families, donors, researchers, and medical personnel, among others. It also reflects our growing international presence and our welcoming spirit to those families where PMS is not caused by a deletion.

We'd like to thank our many corporate and family sponsors, including **Platinum sponsors Cedar Street Advisors and Brookfield Renewable Energy**.

We'd like to hear your thoughts about the conference. Please complete the online survey that was e-mailed to everyone who attended. Based on your input we'll be able to make 2016 an even better experience!