FOR IMMEDIATE RELEASE
September 22, 2023
Contact:

International Phelan-McDermid Syndrome Awareness Day is Oct. 22nd

OSPREY, FLA. – The Fifth Annual International Phelan-McDermid Syndrome Awareness Day will be celebrated on October 22nd to raise awareness of Phelan-McDermid syndrome (PMS), a rare genetic condition affecting more than 3,500 people worldwide. It's estimated that 1 percent of people with autism have PMS, but many are undiagnosed.

Families and advocates of those with PMS around the world are raising awareness the week of October 22 by "shining green" on homes and local landmarks. Participants are encouraged to share their stories on social media should using the hashtag #PMSAD.

Phelan-McDermid syndrome is caused by a deletion or variation of the long arm of chromosome 22 in the 22q13 region, most often including the SHANK3 gene, or a disease-causing mutation of the SHANK3 gene. The diagnosis of Phelan-McDermid syndrome is made definitively by genetic testing. This minor change in one chromosome can have a devastating health impact on those affected by the syndrome.

There is no cure or treatment for PMS. Though improvements can be attained through pharmacologic treatment of associated medical conditions, aggressive speech, occupational and physical therapy and intense education programming, people with PMS are rarely ever able to live independently into adulthood. PMS symptoms vary, but they often cause a wide range of medical, intellectual, and behavioral challenges, delayed or absent speech, epilepsy, low muscle tone and motor delays. Many individuals experience regressions and develop neuropsychiatric issues later in life.

Raising awareness will help improve support services, basic research, drug development, and policymaking related to the syndrome. Increased funding for PMS research will improve health outcomes and improve the quality of life for individuals living with the PMS.

For more information about Phelan-McDermid syndrome, go to (What is PMS? page)

About the Phelan-McDermid Syndrome Foundation go to www.pmsf.org or contact the PMS Foundation at info@pmsf.org.

The Phelan-McDermid Syndrome Foundation is a 501(c)(3) nonprofit organization founded in 1998 and based in Osprey, Fla. It supports families and medical professionals by raising awareness about the syndrome and building alliances that accelerate research. The PMS Foundation is the leading organization worldwide supporting awareness and research into this rare genetic syndrome.