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THE GENERAL ASSEMBLY OF PENNSYLVANIA

SENATE RESOLUTION

No. 419 Session of 2015

INTRODUCED BY BOSCOLA, FARNESE, SABATINA, TEPLITZ, HUTCHINSON AND RAFFERTY, AUGUST 30, 2016

INTRODUCED AND ADOPTED, AUGUST 30, 2016

A RESOLUTION

Designating September 18, 2016, as "Pitt Hopkins Syndrome Awareness Day" in Pennsylvania.

WHEREAS, Pitt Hopkins syndrome is a rare and severe neurological disorder caused by a spontaneous mutation on the 18th chromosome; and

WHEREAS, Pitt Hopkins syndrome is characterized by developmental delays, moderate to severe intellectual disability, breathing problems, epilepsy or recurrent seizures, gastrointestinal issues and distinctive facial features; and

WHEREAS, Most affected individuals do not learn to walk unassisted and do not develop speech, but not all symptoms need to be present to warrant genetic testing in individuals; and

WHEREAS, Pitt Hopkins syndrome was first described by two physicians in 1978, however, until recently, the diagnosis was based on a person's medical history and a physical examination; and

WHEREAS, New technology has allowed scientists to identify the genetic cause and now the diagnosis is usually based on the

results of microarray analysis which can be performed on a blood sample; and

WHEREAS, In 2007, the connection to the 18th chromosome was established when the TCF4 transcription factor located on chromosome 18 was discovered to play an essential role in the development of the nervous system and the brain, and any functional deficiency in development affects how a child develops over time; and

WHEREAS, Approximately 550 people worldwide have been diagnosed with Pitt Hopkins syndrome, however, given the fact that the discovery of the gene happened several years ago, there are likely many others out there that remain undiagnosed; and

WHEREAS, Pitt Hopkins syndrome occurs in both males and females and is not limited to a specific ethnic background; and

WHEREAS, This condition is inherited in an autosomal dominant pattern, meaning one copy of the altered gene in each cell is sufficient to cause the disorder, but most cases result from new mutations in the gene and occur in people with no history of the disorder in their family; and

WHEREAS, Given its rarity, online support groups play a pivotal role for individuals, families and their loved ones to connect with each other and increase awareness; and

WHEREAS, The Pitt Hopkins Research Foundation celebrates September 18, 2016, as the fourth annual International Pitt Hopkins Syndrome Day to raise awareness and funding for Pitt Hopkins research; and

WHEREAS, This date was chosen to bring attention to the fact that it is a disorder caused by a spontaneous mutation on the 18th chromosome; and

WHEREAS, The Pitt Hopkins Research Foundation is an all-

volunteer organization of families and professionals dedicated to finding a cure for Pitt Hopkins syndrome through the funding of research, education and advocacy; and

WHEREAS, The foundation's goal is to bring practical treatment into current medical practice; therefore be it

RESOLVED, That the Senate designate September 18, 2016, as "Pitt Hopkins Syndrome Awareness Day" in Pennsylvania in recognition of the needs of individuals with Pitt Hopkins syndrome and urge all residents to support the need for awareness and education regarding Pitt Hopkins syndrome in order to assist those individuals and families who deal with this syndrome every day of their lives.