THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE RESOLUTION

_{No.} 917

Session of 2020

INTRODUCED BY HICKERNELL, EMRICK, JONES, KORTZ, LONGIETTI, MENTZER, MILLARD, MURT, NEILSON, READSHAW, ROZZI, RYAN, SAYLOR, SONNEY, TOMLINSON AND ZIMMERMAN, JUNE 23, 2020

REFERRED TO COMMITTEE ON HEALTH, JUNE 23, 2020

A RESOLUTION

- Designating the month of July 2020 as "MECP2 Duplication Syndrome Awareness Month" in Pennsylvania.
- 3 WHEREAS, MECP2 duplication syndrome is usually caused by
- 4 duplication of DNA on the Xq28 region of the chromosomes; and
- 5 WHEREAS, MECP2 duplication syndrome causes many problems and
- 6 is typically characterized by hypotonia, developmental delay,
- 7 intellectual disability, limited or absent speech, constipation,
- 8 reflux, ataxia, progressive spasticity, stereotyped movements of
- 9 hands, teeth grinding, recurrent respiratory infections,
- 10 epilepsy and developmental regression in boys; and
- 11 WHEREAS, Some boys have large duplications which include many
- 12 other genes, and the full extent of phenotypes due to
- 13 duplication of other genes is not completely understood at this
- 14 time; and
- 15 WHEREAS, Recent studies have shown a link between MECP2
- 16 duplication syndrome and autism, and most boys with this
- 17 syndrome do have distinct features of autism; and

- 1 WHEREAS, Patients with MECP2 duplication syndrome are highly
- 2 responsive to proper stimulation such as schooling, exercise,
- 3 music, age-appropriate social interactions and related
- 4 activities; and
- 5 WHEREAS, MECP2 duplication syndrome was not formally
- 6 recognized until 2005; and
- WHEREAS, MECP2 duplication syndrome is most commonly
- 8 inherited in an X-linked manner, with affected males most
- 9 commonly inheriting the MECP2 duplication from the carrier
- 10 mother, but spontaneous duplications have been reported; and
- 11 WHEREAS, To date, no cases of a father transmitting the
- 12 duplication have been reported; and
- 13 WHEREAS, MECP2 duplication syndrome research, which has shown
- 14 that symptoms of the disorder can be reversed in mice using a
- 15 small antisense oligonucleotide, gives hope for the future
- 16 development of effective treatments for humans with this
- 17 disorder; and
- 18 WHEREAS, Clinical readiness studies are ongoing to identify
- 19 ways to monitor response to therapy and ensure safety as a
- 20 preparation for future clinical trials; therefore be it
- 21 RESOLVED, That the House of Representatives designate the
- 22 month of July 2020 as "MECP2 Duplication Syndrome Awareness
- 23 Month" in Pennsylvania.