

THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE RESOLUTION

No. 662 Session of
2018

INTRODUCED BY D. COSTA, HENNESSEY, RYAN, NEILSON, READSHAW,
THOMAS, TOEPEL, KINSEY, BERNSTINE, MURT, CALTAGIRONE,
SCHWEYER, J. McNEILL, WARD, MILLARD, DIGIROLAMO, JOZWIAK AND
ZIMMERMAN, JANUARY 29, 2018

INTRODUCED AS NONCONTROVERSIAL RESOLUTION UNDER RULE 35,
JANUARY 29, 2018

A RESOLUTION

1 Recognizing individuals living with maple syrup urine disease
2 and raising awareness of the disease in Pennsylvania.

3 WHEREAS, Maple syrup urine disease (MSUD), also called
4 branched-chain ketoaciduria, is an inherited disease in which
5 the body is unable to process certain amino acids, causing a
6 harmful buildup of substances in the blood and urine; and

7 WHEREAS, MSUD is characterized by a distinctive sweet odor of
8 the affected individual's urine, which is reminiscent of maple
9 syrup; and

10 WHEREAS, Symptoms usually appear within the first few days or
11 weeks after birth and may include lethargy, irritability and
12 poor feeding; and

13 WHEREAS, MSUD can lead to more severe symptoms, including
14 progressive brain damage, coma and even death; and

15 WHEREAS, MSUD takes five distinct clinical variants,
16 including classic MSUD, intermediate MSUD, intermittent MSUD,

1 thiamine-responsive MSUD and E3-deficient MSUD with lactic
2 acidosis, which can be distinguished by age of onset, severity
3 of clinical symptoms and response to treatment; and

4 WHEREAS, Worldwide, MSUD occurs in approximately 1 in every
5 185,000 live births, with a high percentage of cases occurring
6 in the Amish and Mennonite communities of central Pennsylvania;
7 and

8 WHEREAS, MSUD is identified in infants through newborn
9 screening programs; and

10 WHEREAS, A diagnosis may also be made based upon symptomatic
11 findings, urine analysis or blood analysis; and

12 WHEREAS, MSUD can be managed through a lifelong adherence to
13 a protein-restricted diet and medical intervention for treatment
14 of metabolic crisis, including dialysis or hemofiltration; and

15 WHEREAS, Families with a child suffering from MSUD can face
16 substantial difficulties finding treatment due to the rarity and
17 lack of awareness of the disease; and

18 WHEREAS, Idario Santos and his family, who are residents of
19 Pittsburgh, Pennsylvania, moved from Brazil in 2005 for Idario's
20 son Artur to receive treatment for his life-threatening
21 diagnosis of MSUD; and

22 WHEREAS, To bring awareness to MSUD and document the
23 difficulties that Idario's family and so many others with the
24 disease have encountered, Idario chronicled the journey to find
25 treatment for his son's diagnosis of MSUD in his book *Sweet*
26 *Odyssey*; and

27 WHEREAS, The importance of an early diagnosis of MSUD and the
28 correct treatment practices can greatly reduce a child's risk of
29 serious complications that may arise from this disease;
30 therefore be it

1 RESOLVED, That the House of Representatives recognize
2 individuals living with maple syrup urine disease and raise
3 awareness of the disease in Pennsylvania; and be it further
4 RESOLVED, That the House of Representatives support efforts
5 to diagnose children with MSUD at an early age and promote
6 initiatives to bring awareness to this life-threatening disease.