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THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE RESOLUTION

No. 332 Session of 2015

INTRODUCED BY BAKER, GINGRICH, READSHAW, DiGIROLAMO, SCHLOSSBERG, HENNESSEY, MILLARD, V. BROWN, COHEN, DUSH, SONNEY, BROWNLEE, KINSEY, WHEELAND, MURT, LONGIETTI, THOMAS, DONATUCCI, CAUSER, McNEILL, ROSS, MARSICO, WARD, WATSON, PICKETT, PAYNE, MAHONEY, FARINA, VEREB, GOODMAN AND D. COSTA, MAY 11, 2015

INTRODUCED AS NONCONTROVERSIAL RESOLUTION UNDER RULE 35, MAY 11, 2015

A RESOLUTION

Designating the week of May 17 through 23, 2015, as "22q11.2 Deletion Syndrome Awareness Week" in Pennsylvania.

WHEREAS, 22q11.2 Deletion Syndrome, also known as DiGeorge syndrome, Velo-cardio-facial syndrome and conotruncal anomaly face syndrome, among others, is a congenital defect in which a small part of chromosome 22 is missing from the time of conception; and

WHEREAS, 22q11.2 Deletion Syndrome is the most commonly known chromosomal microdeletion syndrome, affecting an estimated 1 in 2,000 to 4,000 live births, yet it is often undiagnosed, especially in adults; and

WHEREAS, For a parent who has 22q11.2 Deletion Syndrome, there is a 50% chance of passing a copy of chromosome 22 with that deletion with each pregnancy; and

WHEREAS, 22q11.2 Deletion Syndrome can affect almost every

system in the body and cause a wide range of health problems; and

WHEREAS, Clinical variability, lack of knowledge and, most importantly, the unfamiliarity of clinicians with this syndrome, all contribute to delayed or missed detection, often resulting in avoidable medical complications and inadequate early interventions; and

WHEREAS, Though not all people with 22q11.2 Deletion Syndrome are affected in the same way, key characteristics of the syndrome include combinations and varying degrees of heart defects, feeding and gastrointestinal difficulties, immune system deficits, growth delay, palate differences, kidney problems, hearing loss, low calcium and other endocrine issues, cognitive and speech delay and behavioral, emotional and psychiatric differences such as ADHD, autism and anxiety; and

WHEREAS, Most people with 22q11.2 Deletion Syndrome are missing a sequence of about 1.5 to 3 million DNA building blocks on one copy of chromosome 22 in each cell, which amounts to about 40 to 50 genes, resulting in multisystem involvement, with the cardiovascular system being the most notable system affected; and

WHEREAS, Symptoms of the disorder do not consistently appear to be related to the amount of genetic material lost in the chromosomal deletion and therefore, there may be a great deal of variation in symptoms among affected people; and

WHEREAS, The symptoms range from serious heart defects to immune deficiency and developmental delays and while not all issues are life-threatening, they all affect the quality of life of the individual; and

WHEREAS, The 22q11.2 deletion is most often not inherited

from either parent and does not usually run in families, and only about 10% of children with the deletion have a parent who is also affected; and

WHEREAS, If the condition is suspected, a blood test to detect the deletion can be performed; and

WHEREAS, Clinic visits and hospital admissions present opportunities to diagnose 22q11.2 Deletion Syndrome, but without caregiver knowledge of this syndrome and its characteristics, patients will not receive the correct diagnosis; and

WHEREAS, Although there is no cure for chromosome 22q11.2 deletion, there are many therapies and medical interventions available to treat the symptoms and the earlier the symptoms are detected, the more doctors can do to help; and

WHEREAS, The 22q and You Center at The Children's Hospital of Philadelphia is an internationally recognized leader in the diagnosis and treatment of children with chromosome 22q11.2 deletion; and

WHEREAS, The Children's Hospital of Philadelphia has evaluated more than 1,200 patients with chromosome 22q11.2 deletion and is the largest program in the country specializing in the condition; and

WHEREAS, With broader awareness of this complex syndrome, the likelihood of a correct diagnosis can be substantially increased; therefore be it

Resolved, That the House of Representatives designate the week of May 17 through 23, 2015, as "22q11.2 Deletion Syndrome Awareness Week" and urge its observance throughout this Commonwealth for the benefit of all individuals living with 22q11.2 Deletion Syndrome and their families.