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THE GENERAL ASSEMBLY OF PENNSYLVANIA

# HOUSE RESOLUTION

No. 308 Session of 2013

INTRODUCED BY TOOHIL, COHEN, HENNESSEY, DiGIROLAMO, BAKER, ROZZI, MIRABITO, KILLION, MILLARD, MAJOR, DONATUCCI, RAPP, GROVE, READSHAW, GRELL, V. BROWN, YOUNGBLOOD, ROCK, KORTZ, ROSS, BENNINGHOFF, HESS, GINGRICH, DENLINGER, FLECK, QUINN, GILLEN, DEASY, BROOKS AND MAHONEY, MAY 8, 2013

INTRODUCED AS NONCONTROVERSIAL RESOLUTION UNDER RULE 35, MAY 8, 2013

A RESOLUTION

Designating the month of June 2013 as "Cockayne Syndrome Awareness Month" in Pennsylvania and commending the work of the Share and Care Cockayne Syndrome Network for raising awareness of this disease.

WHEREAS, Cockayne Syndrome, or CS, is a rare genetic disorder affecting a small number of children worldwide, but its impact on involved families is significant emotionally and economically; and

WHEREAS, Edward Alfred Cockayne (1880-1956), after whom this disease is named, was a London physician who concentrated particularly on hereditary diseases of children; and

WHEREAS, CS is inherited in an autosomal recessive pattern; therefore, in order for a child to be affected by CS, he or she must inherit a mutation in the same CS gene from both parents; and

WHEREAS, While parents, who are carriers of a single CS gene mutation, remain healthy after the birth of a child with CS,

they have a one in four, or 25%, chance of having a second or successive child with CS; and

WHEREAS, The symptoms of CS vary significantly, especially with regard to the age of onset and rate of progression, and include social, jovial personalities; sunburning easily; progeria (premature aging); shortened life span; microcephaly; neurodevelopment delay; short stature (height lower than the 5th percentile for others in the age group); contractures; unsteady gait; spasticity; rounded back; deep-set eyes and a small, slender, straight nose; dental caries (cavities); retinopathy and cataracts; hearing loss; poor circulation (cold hands and feet); low body temperature; feeding problems; sleeping with eyes open; tremors; white matter abnormalities; basal ganglia calcifications; liver abnormalities; elevated liver enzymes; hypertension; and severe itchiness; and

WHEREAS, The resulting spectrum of severity can be loosely divided into three "types" of CS:

(1) CS Type I is characterized by normal prenatal growth with the onset of growth and developmental abnormalities around one year of age. The typical lifespan is 10 to 20 years of age.

(2) CS Type II is characterized by growth failure and other abnormalities at birth, with little or no postnatal neurological development. The typical lifespan is up to seven years of age.

(3) CS Type III is characterized by a later onset, lesser symptoms and a slower rate of progression. The expected lifespan is unclear, but can extend to 40 or 50 years of age;

and

WHEREAS, Some individuals have combined features of CS and Xeroderma Pigmentosum, which is characterized by a wide range of skin changes from mild freckling to skin cancer on areas exposed to sunlight; and

WHEREAS, No specific treatment currently exists for CS, and patients are treated according to the symptoms they have, with physical, occupational, speech, vision and hearing therapies being beneficial; and

WHEREAS, It is important to those affected by CS to raise awareness of this disease so that they find social and medical support easily, and the Share and Care Cockayne Syndrome Network provides information and support for those afflicted; therefore be it

RESOLVED, That the House of Representatives designate the month of June 2013 as "Cockayne Syndrome Awareness Month" in Pennsylvania and commend the work of the Share and Care Cockayne Syndrome Network for raising awareness of this disease.