
THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE RESOLUTION

No. 954 Session of
2015

INTRODUCED BY TOOHL, BAKER, COHEN, D. COSTA, DIGIROLAMO,
DONATUCCI, DUSH, GILLEN, GOODMAN, HENNESSEY, KINSEY,
KIRKLAND, LONGIETTI, MACKENZIE, MAJOR, MARSICO, McNEILL,
MILLARD, MILNE, MURT, NEILSON, OBERLANDER, READSHAW, ROSS,
ROZZI, SONNEY, WARD, WHEELAND AND YOUNGBLOOD, JUNE 29, 2016

INTRODUCED AS NONCONTROVERSIAL RESOLUTION UNDER RULE 35,
JUNE 29, 2016

A RESOLUTION

1 Designating the month of June 2016 as "Cockayne Syndrome
2 Awareness Month" in Pennsylvania and commending the work of
3 the Share and Care Cockayne Syndrome Network for raising
4 awareness of this disorder.

5 WHEREAS, Cockayne syndrome (CS) is a rare genetic disorder
6 that affects a small number of children worldwide, but its
7 impact on affected families is emotionally and economically
8 significant; and

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

12 WHEREAS, CS is inherited in an autosomal recessive pattern:
13 in order for a child to be affected by CS, the child must
14 inherit a mutation in the same CS gene from both parents; and

15 WHEREAS, While parents who are carriers of a single CS gene
16 mutation remain healthy, after the birth of a child with CS they
17 have a 1 in 4, or 25%, chance of having a second or successive

1 child with CS; and

2 WHEREAS, The symptoms of CS vary significantly, especially
3 with regard to the age of onset and rate of progression, and
4 include social and jovial personalities, sunburning easily,
5 progeria (premature aging), shortened lifespan and microcephaly;
6 and

7 WHEREAS, Neurodevelopment delay, short stature (height lower
8 than the 5th percentile for others in the age group),
9 contractures, unsteady gait, spasticity, rounded back, deep-set
10 eyes and a small, slender, straight nose are other symptoms and
11 features associated with CS; and

12 WHEREAS, Dental caries (cavities), retinopathy, cataracts,
13 hearing loss, poor circulation (cold hands and feet), low body
14 temperature, feeding problems, sleeping with eyes open, tremors,
15 white matter abnormalities, basal ganglia calcifications, liver
16 abnormalities, elevated liver enzymes, hypertension and severe
17 itchiness are other characteristics of CS; and

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

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

24 (2) CS Type II is characterized by growth failure and
25 other abnormalities at birth, with little or no postnatal
26 neurological development. The typical lifespan is up to 7
27 years of age.

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

1 and

2 WHEREAS, Some individuals have combined features of CS and
3 xeroderma pigmentosum, which is characterized by a wide range of
4 skin changes from mild freckling to skin cancer on areas exposed
5 to sunlight; and

6 WHEREAS, No specific treatment currently exists for CS; and

7 WHEREAS, Patients are treated according to the symptoms they
8 have, with physical, occupational, speech, vision and hearing
9 therapies being beneficial; and

10 WHEREAS, It is important to persons with CS and their
11 families to raise awareness of this disorder so that they may
12 find social and medical support more easily; and

13 WHEREAS, The Share and Care Cockayne Syndrome Network
14 provides information and support for affected individuals and
15 their families; therefore be it

16 RESOLVED, That the House of Representatives designate the
17 month of June 2016 as "Cockayne Syndrome Awareness Month" in
18 Pennsylvania and commend the work of the Share and Care Cockayne
19 Syndrome Network for raising awareness of this disorder.