

Costello syndrome is an exceedingly rare genetics syndrome first reported in 1977 by Dr. Jack Costello, a geneticist in New Zealand. With an estimated 250 people in the world, the incidence is 1:24 million, or fewer than 10 babies born with the syndrome per year worldwide. Researchers have recently identified Costello syndrome associated with mutations on the HRAS gene.

Features

Major Features

- ◆ Dysphagia / Feeding difficulty / Gastrostomy tube (g-tube) (95%)
- ◆ Postnatal short stature (97%)
- ◆ Characteristic facial features (98%)
- ◆ Thick lips (95%)
- ◆ Loose skin (94%)
- ◆ Abnormal palmar skin creases (99%)
- ◆ DD (developmental delay) / MR (mental retardation) (100%)

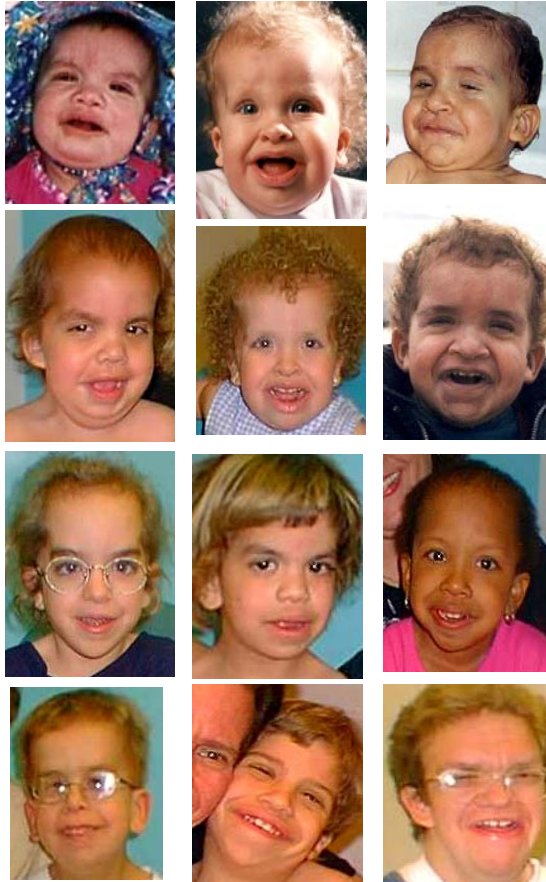
Unique Features

- * Congenital heart problems (65%) including pulmonic stenosis (20%), hypertrophic cardiomyopathy (40%) and atrial tachycardia (30%)
- * Benign (44%) and malignant tumors (16%)
- * Characteristic facial features with large mouth (78%)
- ◆ Stretchy skin with hyperpigmentation
- ◆ Kyphoscoliosis
- ◆ Engaging personality
- ◆ Curly hair
- ◆ Normal head circumference

Other Features

- ◆ Polyhydramnios (62%)
- ◆ Birth weight >50%
- ◆ Hernias (50%)
- ◆ Vision problems - ptosis and strabismus

- * When added to Major Features will greatly increase the specificity of diagnosis



Life-threatening complications

- ◆ Cardiac arrhythmia
- ◆ Hypertrophic cardiomyopathy
- ◆ Malignancy



Natural History

Infants (<1 year)

- Polyhydramnios
- Fetal distress
- C-section (Cesarean section)
- LGA (large for gestational age)
- Feeding problems and hypotonia
- Gastrostomy tube (g-tube) in the first 4 years
- FTT (failure to thrive)
- Growth delay in weight and length but with normal head circumference
- Motor delays

Toddlers (1-3 years)

- Distinctive facial features, broad mouth
- Loose, lax skin, soft, deep wrinkles, abnormal creases in palms and soles, stretchy skin, loose joints
- Hernias
- Hypertrophic cardiomyopathy
- Arrhythmia, especially atrial tachycardia
- Strabismus, ptosis
- Malignancy
- DD (developmental delay)

Children (4-12 years)

- Short stature
- Distinctive facial features with broad mouth and thick lips
- Kyphoscoliosis, cervical kyphosis
- Remarkably pleasant, sociable, humorous and easy-going personality

Adolescents/Adults

- Coarse classic facial features
- Thicker, often curly hair
- Nasal fibromata
- Breast papilloma
- Hyperkeratosis, hyperpigmentation
- Short stature
- Skeletal and orthopedic problems
- DD (developmental delay) / MR (mental retardation)