

PROPOSED GUIDELINES

Based on frequencies found in literature review

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

Costello Syndrome Diagnostic Guidelines

Defining the criteria to diagnose a multiple congenital anomaly syndrome takes many years, many patients and remains largely unscientific, often relying on the “gestalt” (overall impression) of a syndrome to make the final diagnosis.

Costello's first reports in 1971 and 1977 were expanded by der Kaloustian (1991), and Martin and Jones (1991). Recent review articles characterize over 100 patients (Hennekam, 2003), and summarize neuro-logical and behavioral issues (Kawame et al., 2003; Axelrad et al., 2004, Del Rue et al., 2003), cardiac complications (Lin et al., 2002), orthopedic problems (Yassir et al., 2003), malignancies (Gripp et al., 2003), and the adult appearance (White et al., 2005).

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WEB LINKS:

Online Mendelian Inheritance in Man:
<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=218040>

International Costello Syndrome Support Group:
www.costellokids.org.uk

Thanks to Angela Lin , MD, Co-Director, Costello Syndrome Professional AdvisoryBoard
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Costello Syndrome



Guidelines for Clinical Diagnosis

Clinical Geneticists have been working for the past 10 years with families and the International Costello Syndrome Support Group (ICSSG) and the Costello Syndrome Family Network (CSFN) to better understand this disorder. For the 4th International Costello Syndrome Conference in St. Louis, Missouri, we presented diagnostic guidelines for Costello syndrome.

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

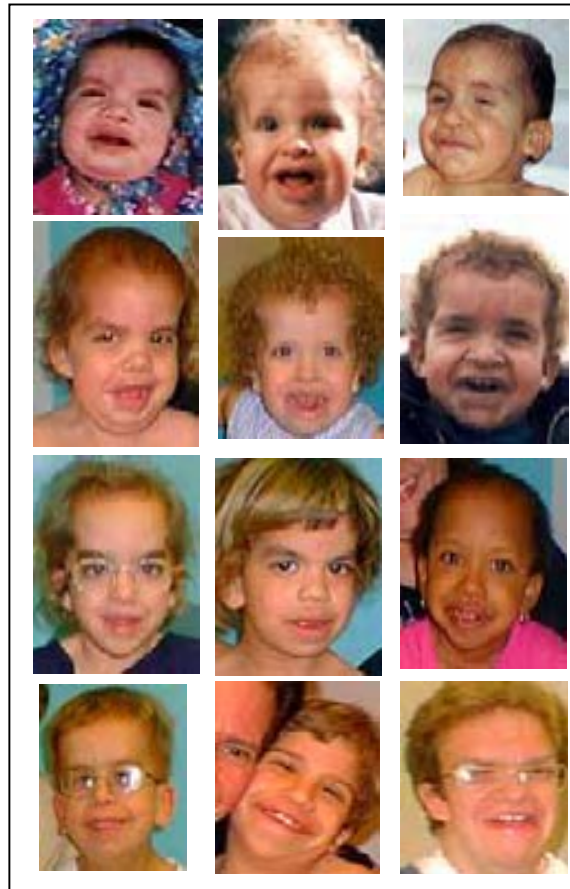
Coarse classic facial features
Thicker, often curly hair
Nasal fibromata
Breast papilloma
Hyperkeratosis, hyperpigmentation
Short stature
Skeletal and orthopedic problems
DD (developmental delay) / MR (mental retardation)



KEY FEATURES

The faces of these individuals, who range in age from infancy to adulthood, illustrate the distinctive appearance of Costello syndrome.

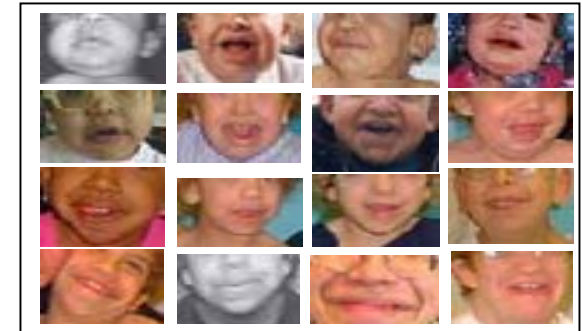
Distinctive facial features



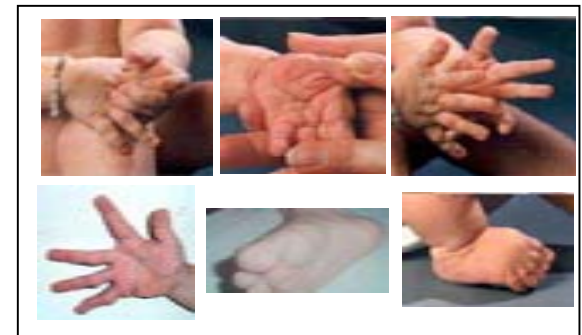
KEY FEATURES

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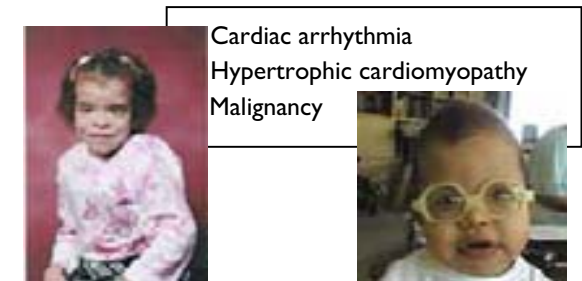
Large mouth and thick lips



Loose skin / deep creases



Life-threatening complications



Cardiac arrhythmia
Hypertrophic cardiomyopathy
Malignancy