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).

--July 2005

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WEB LINKS:
Online Mendelian Inheritance in Man:

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

Thanks to Angela Lin, MD, Co-Director, Costello Syndrome Professional Advisory Board
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**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**

**Large mouth and thick lips**

**Loose skin / deep creases**

**Life-threatening complications**

Photo credits go to the parents of the above children and adults and, for the fourth image on the first panel, Rick Guidotti/Positive Exposure.