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Cancer Prone Disease Section

Mini Review

Costello syndrome

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Identity

Alias

Noonan-like syndrome with nasal papillomata.

Inheritance

The vast majority of cases are sporadic. An increase in mean paternal age has been demonstrated, favouring the hypothesis of dominant de novo mutations, but a microdeletion is an alternative explanation.

Clinics

Note

Costello syndrome is a multiple congenital anomalies/mental retardation syndrome characterised by severe growth abnormalities and a predisposition to develop childhood tumours, especially rhabdomyosarcomas.

Phenotype and clinics

Costello syndrome is characterised by:

- Growth abnormalities: whereas new-born are often macrosomic and macrocephalic they exhibit severe feeding difficulties and failure to thrive during the first months of life, up to two years of age. After this marasmic period, growth velocity is restored but the final height is short.
- Ectodermal abnormalities are characterised by loose and dark-coloured skin, and a predisposition to develop multiple papillomata, which when present are highly suggestive of the diagnosis.
- Mental retardation is usually mild and most patients with CS have a happy, ongoing personality.
- Heart defects are present in one third of patients, either structural defects, hypertrophic cardiomyopathy or dysarrythmia.

Neoplastic risk

Patients with Costello syndrome are prone to develop both benign and malignant tumours. The risk of developing a cancer is up to 15%. Rhabdomyosarcoma, mostly of the embryonic subtype is the tumor the most frequently encountered in CS. Neuroblastomaand bladder cancer (very rare in children) have also been described in several patients.

Treatment

Symptomatic: surgery of congenital heart defects or tumors; tube feeding during the first months.

Prognosis

Apart from mental retardation, the prognosis of patients with Costello syndrome depends mainly on the occurrence of cardiac and/or tumoral complications.

Genes involved and proteins

Note

Unknown.

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