Cancer Prone Disease Section
Short Communication

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Identity

Inheritance
X-linked with heterogeneity; most families map Xq26; one large pedigree maps to Xp22.

Clinics

Phenotype and clinics
Pre-natal and post-natal overgrowth syndrome, similar to Beckwith-Wiedemann syndrome.
Xq26: coarse facies with mandibular overgrowth, cleft palate, heart defects, hernias, supernumerary nipples, renal and skeletal abnormalities.
Xp22: lethal form, multiple anomalies, hydrops fetalis, death within first 8 weeks of life with a neoplastic risk.

Neoplastic risk
Wilms tumor, neuroblastoma during early childhood; one case of hepatocellular carcinoma reported.

Genes involved and proteins

GPC3

Protein
Description: GPC3, an X-linked recessive overgrowth gene, may encode a negative regulator of mesothelial cell growth, based on observation that down-regulation of GPC3 is a common occurrence in malignant mesothelioma.
Function: Proteoglycans are essential cofactors in cell-cell recognition systems, cell-matrix adhesion processes and receptor-growth factor interactions.

References


This article should be referenced as such: