

Simpson-Golabi-Behmel Syndrome

Identity

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

Clinics

Phenotype and clinics Characterized by a wide variety of clinical manifestations including pre-natal and post-natal overgrowth syndrome
SGBS is phenotypically similar to [Beckwith-Wiedemann syndrome \(BWS\)](#) suggesting that at least part of the SGBS phenotype could be due to increased IGF-II signalling.
Xq26: coarse facieses 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.

Neoplastic risk increased risk of embryonal tumors, including [Wilms tumor](#), [neuroblastoma](#) ; one case of [hepatocellular carcinoma](#) reported

Genes involved and Proteins

Gene Name [glypican-3 \(GPC3\)](#)

Location Xq26

Protein

Description GPC3 is highly expressed in embryonal tissues such as the developing intestine and the mesoderm-derived tissues, and its expression is downregulated in most adult tissue, implying a potential role in development. GPC3 is a heparan sulfate proteoglycan (HSPG) that is attached to the cell surface via a glycosyl-phosphatidylinositol (GPI) anchor.

Function HSPGs of the cell surface are highly interactive macromolecules playing various roles in cell migration, proliferation, differentiation and adhesion, and participating in many developmental and pathological processes.

Mutations

Germinal Most cases are caused by deletions of different exons in the GPC3 genes . The exact role of GPC3 in the etiology of SGBS is still unknown. The renal dysplasia observed in both SGBS patients and GPC3-deficient mice could be explained by the participation of GPC3 in the

control of renal branching morphogenesis by modulating the actions of several different growth factors, including BMP2, BMP7 and fibroblast growth factor 7.

External links

[OMIM](#) [312870](#)

[Orphanet](#) [Simpson-Golabi-Behmel syndrome](#)

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