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Gene Section
Short Communication

PTCH (patched homolog)

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Identity

Other names: PTC, but this term was confusing with PTC/PKA.
Location: 9q22.3
Local order: between FACC and XPAC.

DNA/RNA

Description

23 exons, 2 of which are non-coding; 34 kb.

Transcription

Alternate splicing: 3 different 5' termini.

Protein

Description

Glycoprotein; 12 transmembrane domains, 2 extra cellular loops and intracellular N-term and C-term.

Localisation

Transmembrane protein.

Function

Part of a signalling pathway; opposed by the hedgehog gene's product; transmembrane protein, with a probable cell to cell adhesion role; is thought to have a repressive activity on cell proliferation; the recent demonstration of NBCS syndrome (see below) as a chromosome instability syndrome suggests that this protein has a role in DNA maintenance, repair and/or replication.

Homology

Patched (drosophila segment polarity gene).

Mutations

Germinal

Germ-line mutations lead to protein truncation in naevoid basal cell carcinoma syndrome (NBCS) patients (see below).

Somatic

Mutation and allele loss events in basal cell carcinoma, in NBCS and in sporadic basal cell carcinoma are, so far, in accordance with the two-hit model for neoplasia, as is found in retinoblastoma.

Implicated in

Naevoid basal cell carcinoma syndrome (NBCS) or Gorlin syndrome

Disease
Autosomal dominant condition; cancer prone disease (multiple basal cell carcinomas); it is also a chromosome instability syndrome.

Cytogenetics
Spontaneous and induced chromosome instability.

Sporadic basal cell carcinoma

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