PTCH1 (patched homolog 1 (Drosophila))

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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; mRNA: 3.4 kb complete cds; coding sequence: CDS 1 ... 2364.

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

Germline

Germ-line mutations lead to protein truncation in naevoid basal cell carcinoma syndrome (NBCS) patients (see below); mutations types are variable: nucleotide substitutions (missense/nonsense), small deletions, or small insertions mainly, leading to protein truncation; these mutations have been observed in most exons; there is, so far, no hot-spot.

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

Disease
Sporadic basal cell carcinoma, but also benign trichoepithelioma, a tumour often associated with basal cell carcinomas.
**A subset of sporadic primitive neuroectodermal tumours (PNETs) of the central nervous system (cerebral PNETs, medulloblastomas, and desmoplastic medulloblastomas)**

Note

Gorlin syndrome patients have a predisposition for the development of PTENs, while, herein mentioned are sporadic.

**Invasive transitional cell carcinoma of the bladder**

Note

PTCH has been found mutated in rare cases.

**References**


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