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# Gene Section
## Mini Review

# 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

### Germinal

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.

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