Naevoid basal cell carcinoma syndrome (NBCS)
Jean-Loup Huret
Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France
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**Identity**

**Other names:** Gorlin syndrome; Gorlin-Goltz syndrome; Multiple basal cell nevi, odontogenic keratocysts, skeletal anomalies; Fifth phacomatosis; Hydrocephalus, costovertebral dysplasia, sprengel anomaly.

**Inheritance:** autosomal dominant with complete penetrance, but variable expressivity; 40% are de novo mutations; frequency is about 2/10^5 newborns.

**Clinics**

NBCS is a hamartoneoplastic syndrome; it is also a chromosome instability syndrome; hamartomas are localized tissue proliferations with faulty differentiation and mixture of component tissues; they are heritable malformations that have a potential towards neoplasia.

**Phenotype and clinics**

Multiple basal cell carcinomas, appearing as early as 15 yrs;
Jaw keratocysts;
Dyskeratotic palmar/plantar pits;
Skeletal malformations (of ribs, spina bifida occulta...);
Soft tissue calcifications (falx cerebri, ovarian fibroma, diaphragma sellae...);
Facial dysmorphia.

**Neoplastic risk**

Mainly multiple basal cell carcinomas;
Other proliferations (see below) in 60% of patients;
Other malignancies: medulloblastoma, ovarian fibrosarcoma;
Benign proliferations: ovarian fibroma, meningioma, rhabdomyoma, cardiac fibroma.

**Treatment**

Tumour exereses.

**Evolution**

Extensive number of basal cell carcinomas.

**Prognosis**

According to the tumours (basal cell carcinomas are not life threatening, but may be devastating).

**Cytogenetics**

**Inborn condition**

Spontaneous and induced chromosome instability.
Delay in the cell cycle.
NBCS is therefore a chromosome instability syndrome.

**Cancer cytogenetics**

Poorly documented.

**Genes involved and Proteins**

**Complementation groups**

None so far.

**PTCH**

**Location:** in 9q22.3 (between FACC and XPAC!!)

**Protein**

**Description:** glycoprotein with transmembrane domains, extra cellular loops and intracellular domains.

**Localisation:** transmembrane protein.

**Function:** part of a signalling pathway; probable cell to cell adhesion role; may have a repressive activity on cell proliferation; as NBCS syndrome is a chromosome instability syndrome, this protein may have a role in DNA maintenance, repair and/or replication.

**Mutations**

**Germline:** most germ-line mutations in NBCS patients lead to protein truncation, which suggests that developmental anomalies seen in NBCS may be due to haplo-insufficiency; no obvious genotype-phenotype correlations.
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.

References


This article should be referenced as such: