

## **Involvement of Patched (PTCH) Gene in Gorlin Syndrome and Related Disorders: Three Family Cases**

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**Aim.** To find genetic alterations in PTC or other genes of the Shh/PTCH pathway in tumorous and non- tumorous samples from three families and to correlate them with the varying expression of disorders in presented nevoid basal cell carcinoma syndrome (NBCCS) phenotypes.

**Method.** DNA was extracted from archival paraffin-embedded tissues, tumor tissue or peripheral blood leukocytes, and the loss of heterozygosity (LOH) and single strand conformational polymorphism analysis was performed using PCR with primers for polymorphic 9q22.3 markers (D9S196, D9S287, D9S180, D9S127); PTCH exons 3, 6, 8, 13, 15, 16; and smo (smoothed) exon 1. G-banding technique was used for cytogenetic analysis of the peripheral blood lymphocytes.

**Results.** We found a LOH for PTCH in several cases and variability in smo in one case. In one case NBCCS could reasonably be ascribed to hemizygous PTCH inactivation, while in other two families this typical correlation between the syndrome phenotype and the observed genetic alterations could not be established.

**Conclusions.** Further analysis of relatively sparse cases of NBCCS is needed before the symptoms of the syndrome could be convincingly explained by genetic alterations in the Shh/PTCH signalling pathway.

**Key words:** allelic loss; basal cell nevus syndrome; Gorlin syndrome; LOH, loss of heterozygosity; signal pathways