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Nevoid Basal Cell Carcinoma Syndrome

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Description

The Nevoid Basal Cell Carcinoma Syndrome (NBCC), also called as Gorlin syndrome and the basal cell nevus syndrome, is an autosomal dominant disorder that predisposes to basal cell carcinomas of the skin, medulloblastomas, and ovarian fibromas. Its prevalence has been estimated at 1 per 56,000, and 1% to 2% of medulloblastomas and 0.5% of basal cell carcinomas are attributable to the syndrome. Other neoplasms that probably occur to excess include fibrosarcomas, eningiomas, rhabdomyosarcomas, and cardiac fibromas. In addition to benign and malignant tumors, malformations are a striking component. The syndrome is associated with pits of the palms and soles, keratocysts of the jaw and other dental malformations, cleft palate, characteristic coarse facies, strabismus, dysgenesis of the corpus callosum, calcification of the falx cerebri, spina bifida occulta and other spine anomalies, bifid ribs and other rib anomalies, ectopic calcification, mesenteric cysts, macrocephaly, and generalized overgrowth.

The NBCC gene was mapped to chromosome9 and the demonstration that the exact same region is deleted in a high percentage of sporadic basal cell carcinomas and other tumors related to the disorder provided strong evidence that the gene functions as a tumor suppressor. Positional cloning identified a human homologue of Drosophila patched as the gene for this syndrome, and subsequent studies showed that patched is mutated in a high percentage of sporadic basal cell carcinomas. Patched is a negative regulator of the hedgehog pathway, several members of which are known to function as oncogenes in skin and brain tumors. Mutation of patched may be a necessary if not sufficient step in basal cell carcinoma development. Minute basal cell carcinomas are as likely as large tumors to have patched mutations, and all histologic subtypes, whether primary or recurrent, have a high frequency of loss of patched. Tumors with allelic loss on chromosome 9 sometimes show additional areas of loss on other chromosomes, but no tumors have loss on other chromosomes without involvement of chromosome 9. Patched appears to function as a gatekeeper gene in the epidermal cell type from which basal cell carcinomas arise.

In contrast to many other disorders caused by tumor suppressors congenital anomalies are a prominent feature of NBCC. One hypothesis to explain at least some of these anomalies is a two-hit mechanism in which a single fetal or embryonic cell that has lost the normal copy of the gene gives rise to a developmentally abnormal clone. However, other symmetric generalized features of the syndrome (e.g., overgrowth, corpus callosum defects) suggest that loss of just one copy of the NBCC gene exerts an effect on growth and differentiation. The diagnosis of NBCC should be considered in anyone below the age of 30 with a single basal cell carcinoma and in older individuals with multiple basal cell carcinomas. Medulloblastoma, keratocysts of the jaw, and typical skeletal anomalies should raise the suspicion of NBCC regardless of the presence or absence of basal cell carcinomas. Palmar and plantar pits are pathognomonic. DNA-based testing is available for individuals at risk with a family history of NBCC and for sporadic patients as well. The most important follow-up study in affected individuals is dermatologic examination for basal cell carcinomas at intervals of 6 months to 1 year. Yearly dental examinations, with particular attention to the possibility of jaw cysts, are also recommended. Because the frequency of medulloblastoma in this syndrome probably does not exceed 5% and may be as low as 1%, screening studies for this tumor type in children are controversial.