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Clinical Features In Children With Nevoid Basal Cell Carcinoma Syndrome. • 863

Virginia E Kimonis, Alisa M Goldstein, Behram Pastakia, Michael L Yang, Ronald Kase, John J DiGiovanna, Allen E Bale & Sherri J Bale
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Nevoid Basal Cell Carcinoma Syndrome (NBCC), an autosomal dominant disorder linked to 9q22.3-q31, is characterized by multiple basal cell carcinomas (BCCs), keratocysts of the jaw, palmar/plantar pits and calcification of the falx cerebri. Twenty-six families including 105 persons with NBCC participated in a linkage and clinical study at the NIH since 1985. Of these, 30 were children aged from 4 mo. to 19 yr. (mean 11.3 yr., M:F ratio 1:1). On comparing the frequency of the major clinical features, 55% of children and 91% of adults had at least one BCC. The number of BCCs ranged from 1- > 1000 (median 1) & 1-500 (median 31.5) respectively. Jaw cysts occurred in 60% of children and 81% of adults, the no. of cysts ranging from 1-6 (median 3) and 1-28 (median 5) respectively. Lifetable analysis of adults and children indicated that 50% developed their first BCCs by the age of 21.5 yr. and their first jaw cyst by the age of 15 yr. Palmar/plantar pits were observed at approximately the same frequency in children (89%) and adults (87%). Calcification of the falx cerebri was seen in 37% children and 78% adults. On comparison with their unaffected siblings (N=25) features seen exclusively in affected children were frontal bossing (44.8%), hypertelorism (38.5%), Sprengels deformity (17.9%), pectus deformity (14.8%), and cleft lip/palate (6.6%). Sixty percent of affected and 8% of unaffected children had macrocephaly (HC > 2SD) (p < 0.01). Radiological features seen exclusively in affected children were calcification of the tentorium cerebellum (13%), bifid ribs (29%), hemivertebrae (9%), fusion of the vertebral bodies (9%), and flame shaped lucencies of the hand (16%). This study indicates that many of the major features (jaw cysts, BCCs, and falx calcification) develop over time. Therefore a diagnosis can be established in young persons by the presence of palmar pits, and evaluation of the above mentioned clinical and radiological features of NBCC.

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