Session 20: Plenary Session

A54 Slide Session 53: Clinical Genetics, Malformations and Dysmorphology II (continued)

281

Clinical features in 101 persons with Nevoid Basal Cell Carcinoma (NBCC) Syndrome

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NBCC (Gorlin syndrome), an autosomal dominant disorder linked to 9q22.3-q31, is characterized by multiple basal cell carcinomas (BCCs), keratocysts of the jaw, palmar/plantar pits, spine and rib anomalies and calcification of the falx cerebri. We evaluated 46 males and 55 females whose ages ranged from 4 months to 87 yr. Seventy-nine % of whites (69/87) and 33% of African-Americans (4/12) had a history of BCCs or current tumors on examination at the NIH. The total number of BCCs ranged from 1 to >1000 (median 8) and 1 to 3 (median 2) in the two racial groups respectively. Jaw cysts occurred in 74%, with 80% of first cysts developing by the age of 20 yr. The number of cysts ranged from 1 to 28 (median 3). Palmar pits/plantar pits were seen in over 80% of patients. Ovarian fibromas were diagnosed by ultrasound in 9/52 females at a mean age of 30 yr. Medulloblastoma occurred in 4 patients at a mean age of 2.3 yr. Three patients had cleft lip/palate. Physical examination revealed coarse facies in 55%, macrocephaly in 50%, hypertelorism in 43%, frontal bossing in 27%, pectus deformities in 13%, and Sprengels deformity in 11%. Important radiological features included calcification of the falx cerebri in 65%, of the tentorium cerebellum in 23%, bridged sella in 67%, bifid ribs in 23%, vertebral anomalies in 31% and flame shaped lucencies of the hand in 30%. CT/MRI studies of the brain revealed asymetric ventricles in 22% (8/37) and dysgenesis or agenesis of the corpus callosum in 11% (4/37) of patients. Comparing the frequencies of clinical features in affected vs. unaffected relatives showed that shortened fourth metacarpals, scoliosis, and absent/cervical ribs were not found to be increased in NBCC. This study delineates the frequency of clinical and radiological features in NBCC in a large population of JS patients.