Novel mutation of the PATCHED gene in a patient with basal cell naevus syndrome and Tetralogy of Fallot

Basal cell naevus syndrome (BCNS) is an autosomal dominant disease characterised by the presence of basal cell carcinomas, odontogenic keratocysts, palmoplantar pits and calcification in the falx cerebri, and caused by mutational inactivation of the PATCHED (PTCH) gene. We identified a Chinese family with the proband and her mother having features characteristics of BCNS. Interestingly, the proband also had Tetralogy of Fallot and cerebral atrophy, which had not been described in BCNS. To investigate the molecular basis of BCNS in this family, we have performed a mutational analysis of the PTCH gene by denaturing high-performance liquid chromatography. A one-base pair frameshift deletion, 1480delT, leading to truncated patched protein was identified in the proband and her mother. This result is consistent with previous report that mutational inactivation of the PTCH gene is the cause of BCNS in Chinese. (H.K. Dermatol. Venereol. Bull. (2003) 11, 4-8)

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