Interesting case report ECARUCA (2016-06)

A de novo 4.4-Mb microdeletion in 2p24.3/p24.2 in a girl with bilateral hearing impairment, microcephaly, digit abnormalities and Feingold syndrome

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Abstract:
We report a 26-month-old girl with profound hearing impairment, microcephaly, psychomotor retardation, short palpebral fissures, hypertelorism, epicanthic folds, a broad nasal bridge, anteverted nostrils, large low-set ears, micrognathia, brachymesophalangy of the second and the fifth fingers, clinodactyly of bilateral fifth fingers and a wide interdigital space between the first and the second toes, carrying a 4.4 Mb de novo microdeletion of chromosome 2p24.3→ p24.2. This region contains the genes of FAM84A, NBAS, DDX1, MYCNOS and MYCN, of which haploinsufficiency or mutations of the MYCN gene is associated with Feingold syndrome. Brain magnetic resonance imaging revealed cochlear nerve hypoplasia and internal auditory canal stenosis. Temporal bone computed tomography showed abnormal dilation of bilateral vestibular and lateral semicircular canals. The present case provides evidence that haploinsufficiency of MYCN in 2p24.3 deletion in humans can cause structural and functional abnormalities of the inner ear.

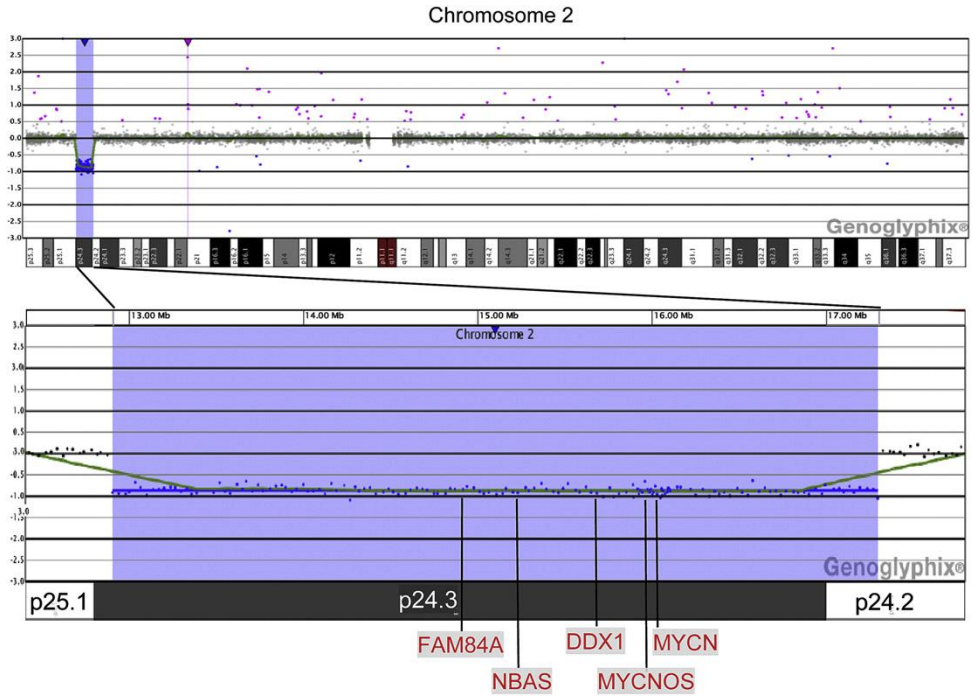

Fig. 1. Array comparative genomic hybridization analysis shows a 4.4-Mb microdeletion of 2p24.3→p24.2 encompassing the genes of FAM84A, NBAS, DDX1, MYCNOS and MYCN.

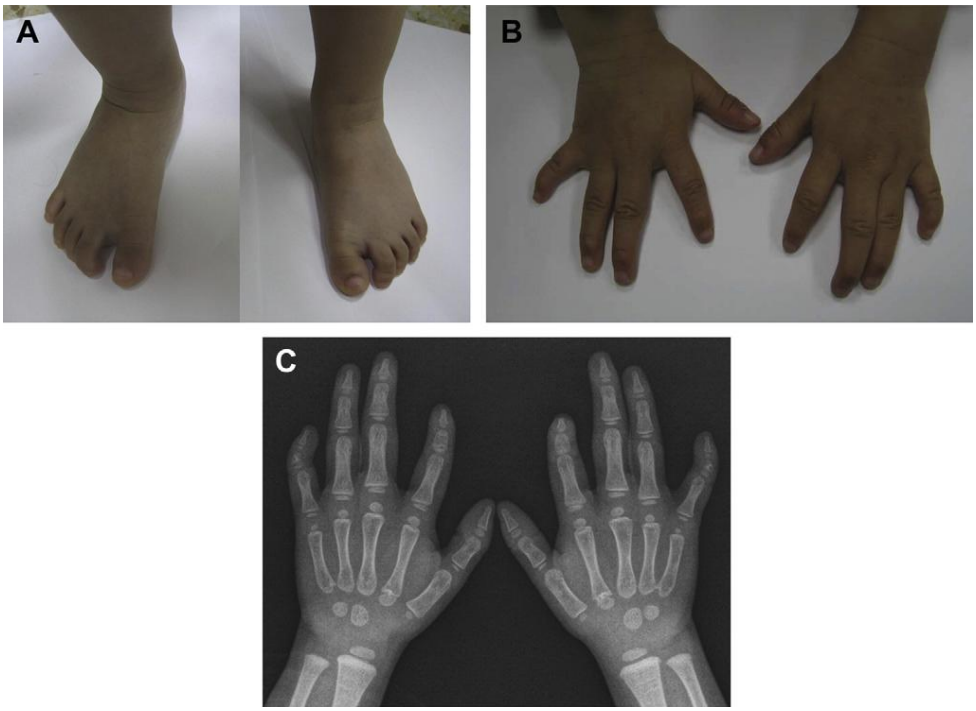

Fig. 2. A) A wide interdigital space between the first and the second toes. (B) and (C) Brachymesophalangy of the second and the fifth fingers, and clinodactyly of bilateral fifth fingers.