Interesting case report ECARUCA (2015-08)

A newborn with a 790 kb chromosome 17p13.3 microduplication presenting with aortic stenosis, microcephaly and dysmorphic facial features - Is cardiac assessment necessary for all patients with 17p13.3 microduplication?

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Abstract:
While deletion of chromosome 17p13.3 (encompassing PAFAH1B1 and YWHAE genes) is known to result in MillereDieker syndrome (OMIM 247200), 17p13.3 microduplication gives rise to a condition commonly associated with developmental delay and autism spectrum disorder. We report a Chinese newborn presenting with dysmorphic features, microcephaly and valvar aortic stenosis, who was confirmed to have a 790 kb microduplication in chromosome 17p13.3 by array comparative genomic hybridization (aCGH). The patient passed away at 4 months of age with presumably life-threatening event associated with his cardiac condition. From literature review, congenital heart diseases of various kinds were identified in up to 20% of patients with 17p13.3 microduplication.We propose cardiac assessment should be part of the comprehensive evaluation of these patients.

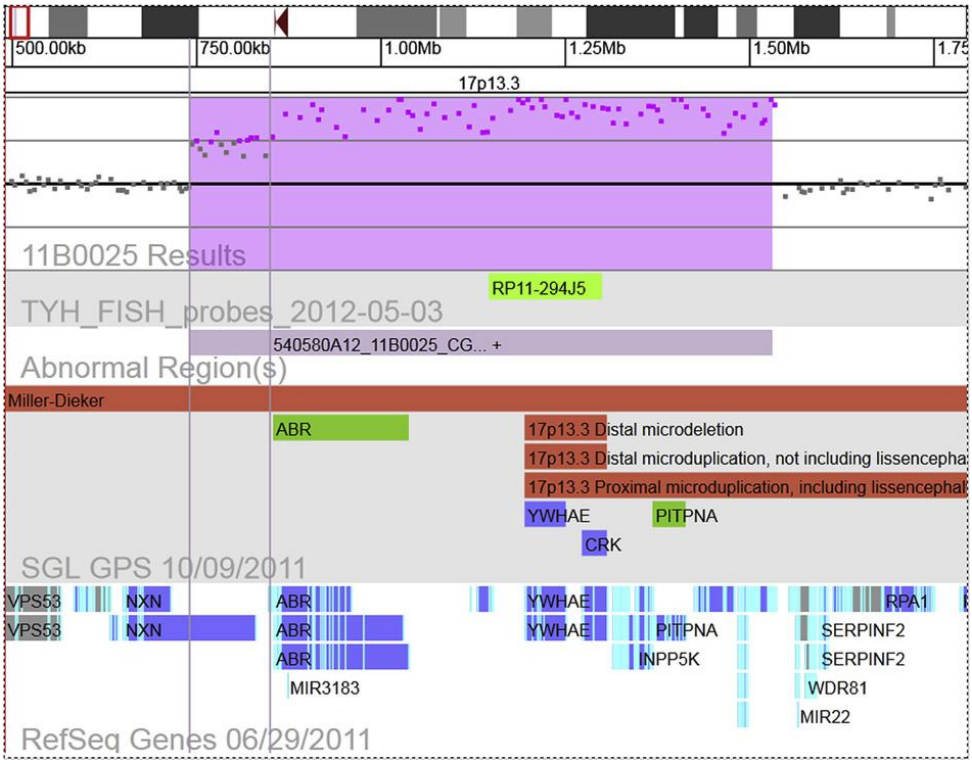

Fig. 1. Result from aCGH showing the duplicated region at chromosome 17p13.3 (pink shaded region). Each dot represents one oligonucleotide probe on the respective chromosome position. The red vertical lines indicated the copy gained region. One-copy gain was found at the probe position of chr17:740,287-849.403, while two-copy gain was at the position of chr17:849,403-1,530,746. The red arrow points to the position of FISH probe. The light blue boxes show the locations of genes in the region.

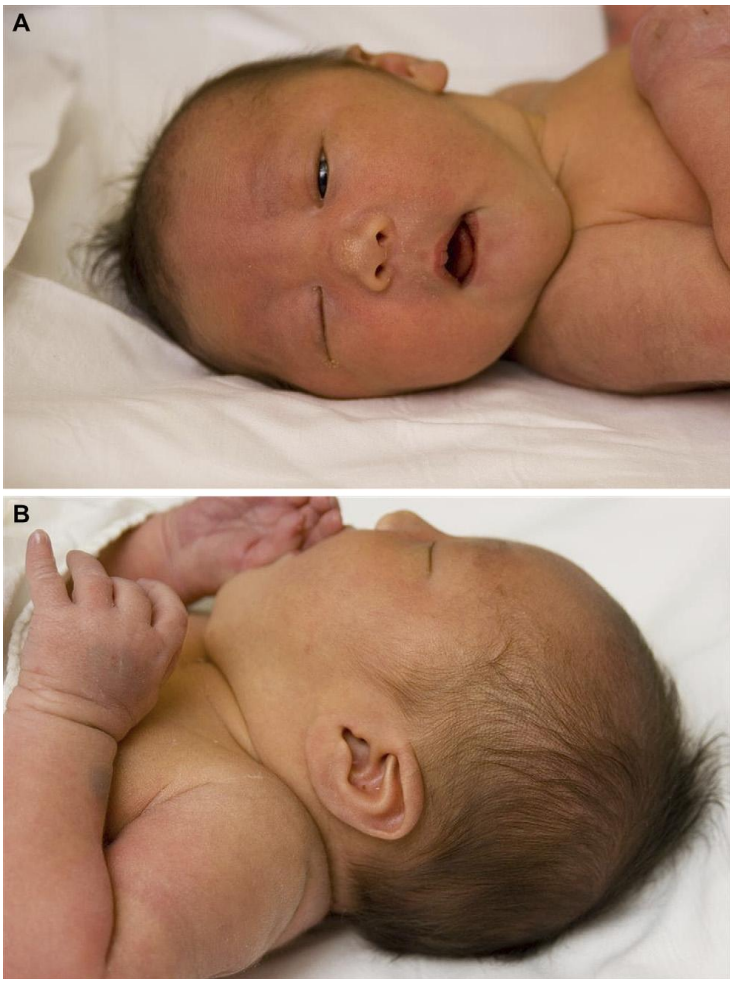

Fig. 2. A) and B) Clinical photographs showing dysmorphic features including flat midface, posteriorly rotated and low set ears, small nose and triangular chin.