

Interesting case report ECARUCA (2016-08)

Duplication of 8q12 encompassing CHD7 is associated with a distinct phenotype but without duane anomaly

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Abstract:

Interstitial duplications of 8q12 encompassing CHD7 have recently been described as a new microduplication syndrome. Three 8q12 duplications have been reported with shared recognizable phenotype: Duane anomaly, developmental delay and dysmorphic facial features. We identified a 2.7 Mb duplication on chromosome 8q12 with SNP-array in a patient with growth delay, congenital heart defects, ear anomalies and torticollis. To our knowledge, this is the smallest duplication reported to date. Our findings support the notion that increased copy number of CHD7 may underlie the phenotype of the 8q12 duplication. Our study together with previous studies suggest that the 8q12 duplication could be defined as a novel syndrome.

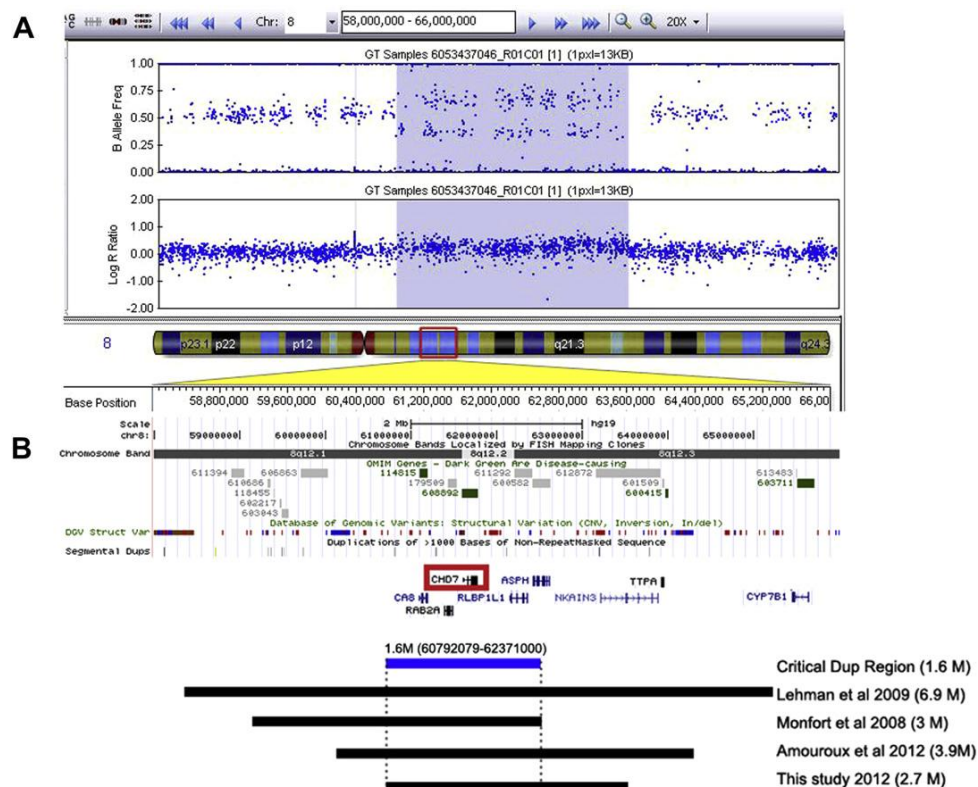


Fig. 1. Illumina SNP-array result of the 8q12 region in the proband. (A) SNP based array shows a 2.7 Mb duplication in 8q12 (Chr8: 60,792,079-63,540,593) (Human GRCh37/hg19 Assembly). B allele frequency and log R ratio are showed in the upper panel; (B). The lower panel shows genes mapped to the duplicated region and the four reported 8q12 duplications in the literature. The smallest region of overlap (1.6 Mb) is indicated. Genomic data have been converted to Hg19.



Fig. 2. Facial features of the proband, lateral (A, C) and frontal (B) view of the patient. B: the patient has a round face, upslanting palpebral fissures and telecanthus, deep set eyes, torticollis (involved the right side), A, C: the patient has a malformation of external right ear.