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De novo microdeletions of chromosome 6q14.1-q14.3 and 6q12.1-q14.1 in two patients with intellectual disability - further delineation of the 6q14 microdeletion syndrome and review of the literature

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Abstract:
Interstitial 6q deletions can cause a variable phenotype depending on the size and location of the deletion. 6q14 deletions have been associated with intellectual disability and a distinct pattern of minor anomalies, including upslanted palpebral fissures with epicanthal folds, a short nose with broad nasal tip, anteverted nares, long philtrum, and thin upper lip. In this study we describe two patients with overlapping 6q14 deletions presenting with developmental delay and characteristic dysmorphism. Molecular karyotyping using array CGH analysis revealed a de novo 8.9 Mb deletion at 6q14.1-q14.3 and a de novo 11.3 Mb deletion at 6q12.1-6q14.1, respectively. We provide a review of the clinical features of twelve other patients with 6q14 deletions detected by array CGH analysis. By assessing all reported data we could not identify a single common region of deletion. Possible candidate genes in 6q14 for intellectual disability might be FILIP1, MYO6, HTR1B, and SNX14.

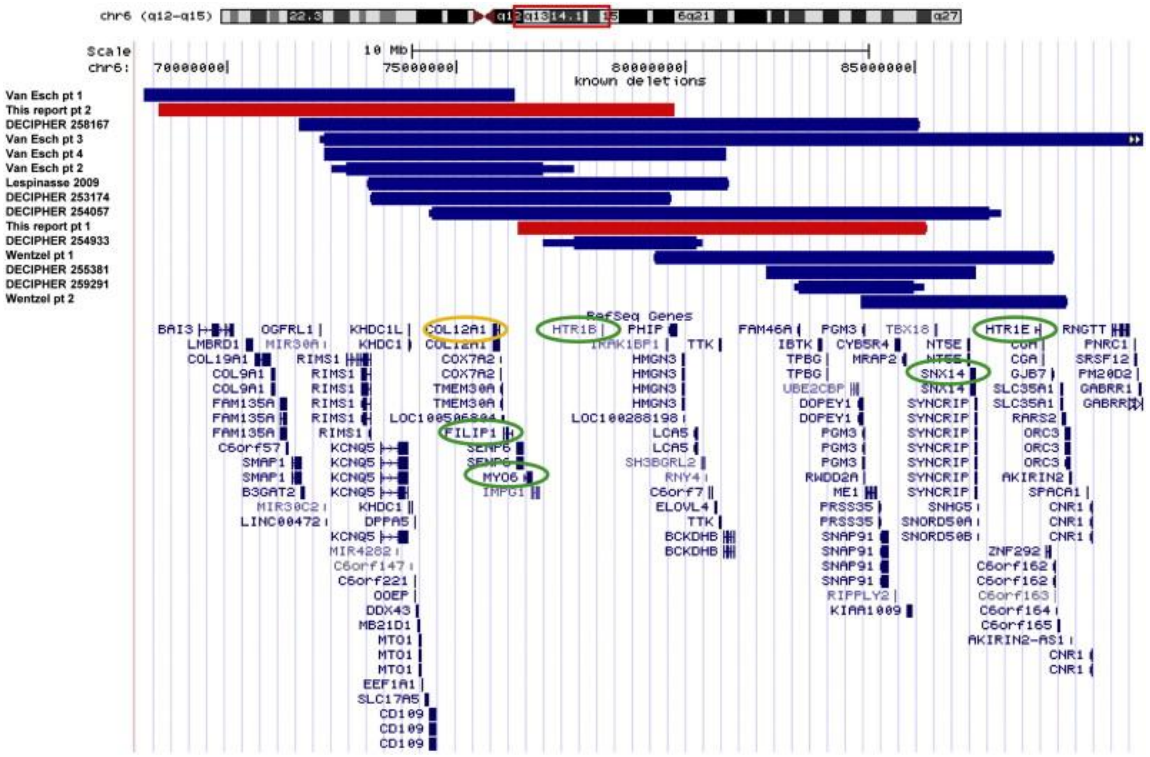

Fig. 1. Comparison of the two cases reported here (marked in red) with thirteen other cases with interstitial 6q deletions and a similar phenotype using the UCSC Genome Browser (GRCh37/hg19). The maximal deletion sizes are marked in narrow bars where available. The deletion of van Esch patient 3 expands beyond the region shown here (white arrows). Potential candidate genes for intellectual disability are circled in green. *COL12A1* as a candidate gene for connective tissue laxity is circled in orange. Abbreviations: pt: patient.

Fig. 2. Patient 1 at the age of one month (left) and one year (middle, right), presenting with facial dysmorphism, including arched eyebrows, synophris, upslanted palpebral fissures with epicanthal folds, small nose, anteverted nares, long philtrum, and thin upper lip.