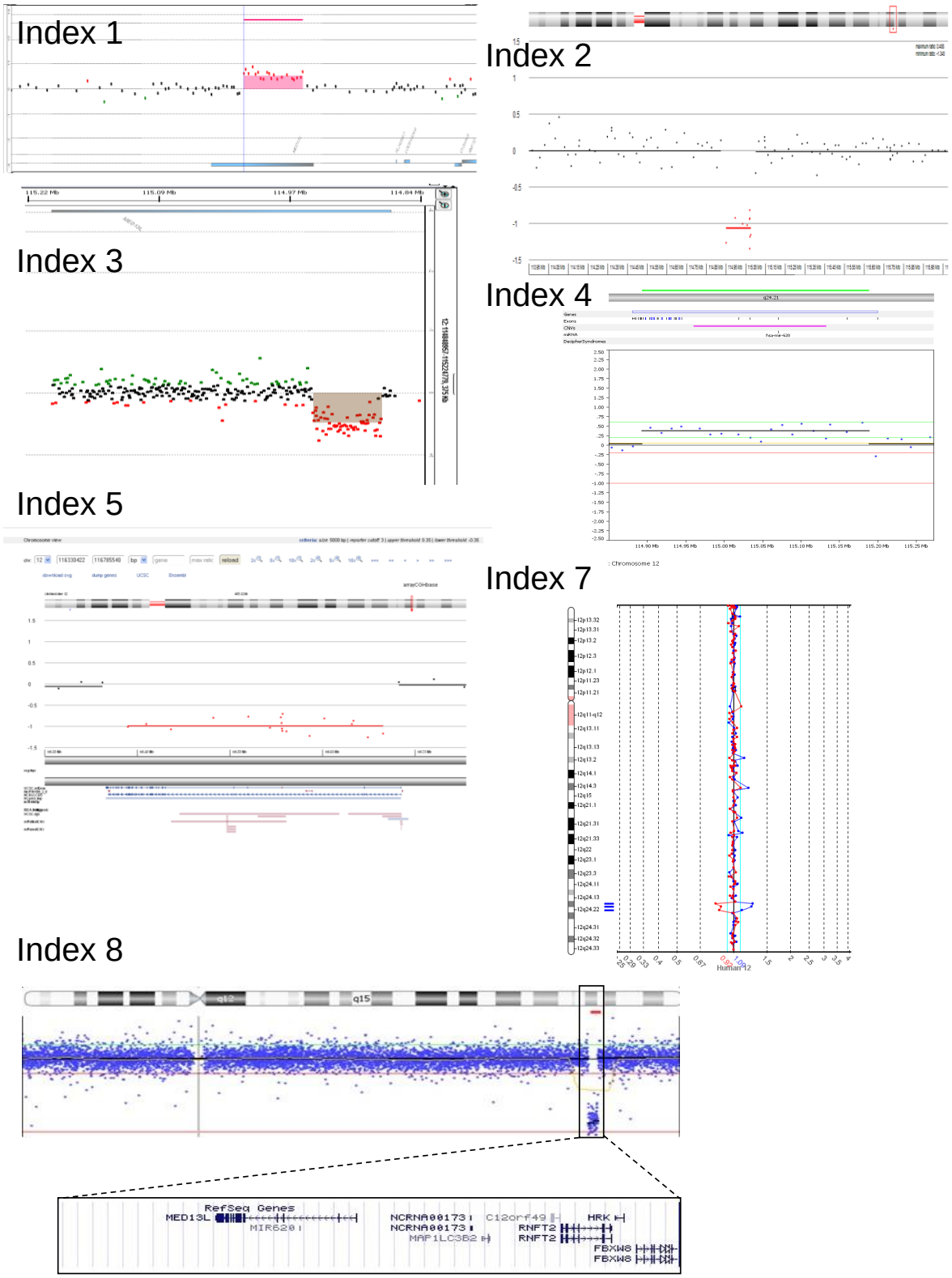
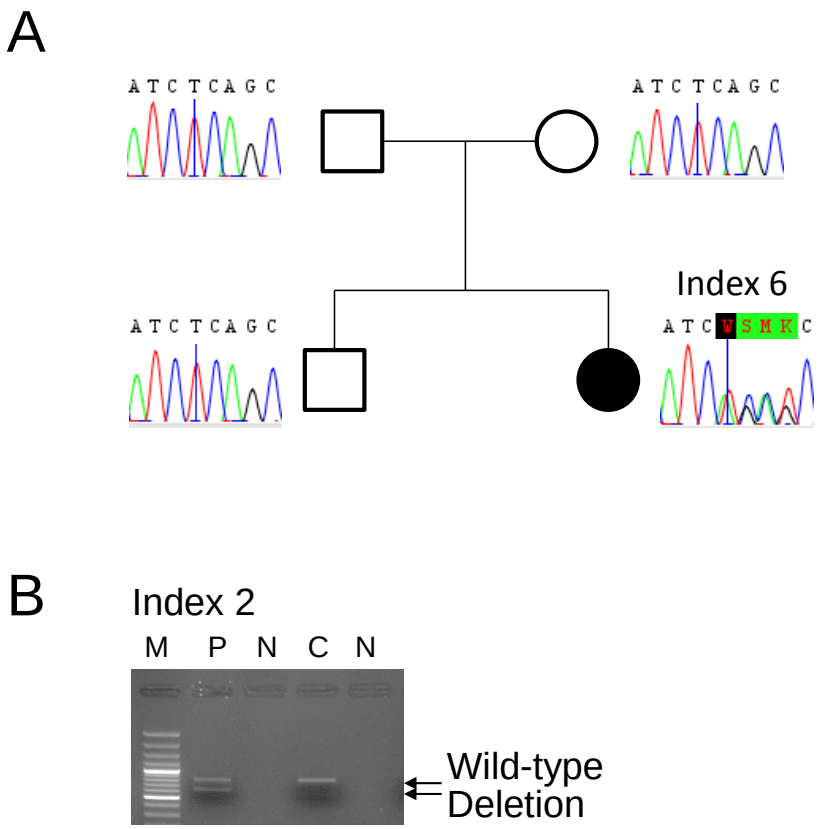
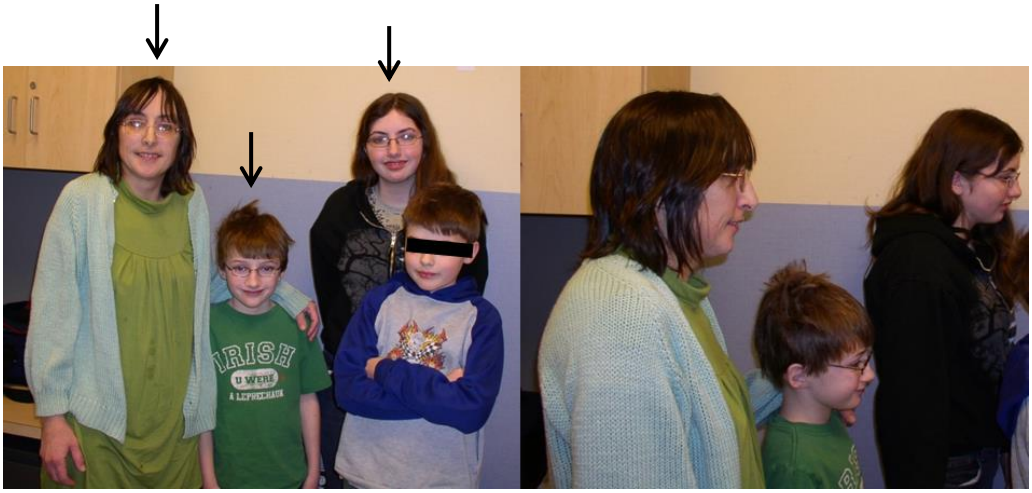
Supplementary online material



Suppl. Figure 1: CGH microarray results for index 1-5, and 7-8.



Suppl. Figure 2: A. Pedigree and Sanger sequencing analysis in family of index 6. Heterozygous mutation in *MED13L* identified by whole exome sequencing is present in the patient but not in her unaffected siblings. **B**. RT-PCR products from index 2 and a control showing two bands in the patient (wild-type and mutant, as confirmed by Sanger sequencing, data not shown) and the wild-type band in the control sample. N, negative control.



Suppl. Fig. 3: Photographs showing index 7 (second from left), his mother and sister who also carry the duplication (arrows indicate carriers of duplication). Affected individuals share a facial appearance consisting of triangular facies, hypertelorism, short philtrum, slightly prominent columella, low-set ears with irregular antihelices, widely spaced teeth, and other features including myopia, and late menarche in both affected women.