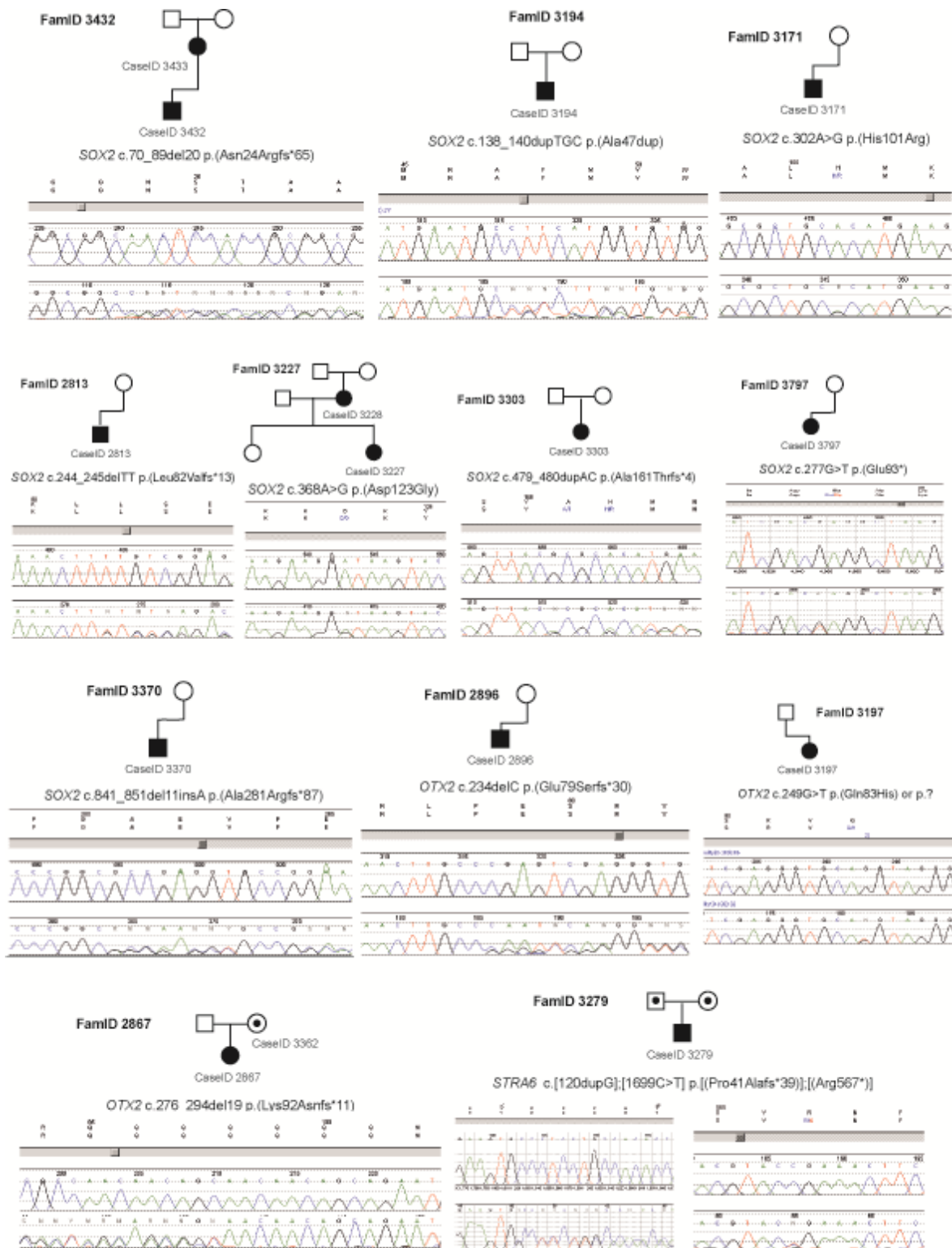


**Supp. Figure S1.** Array CGH identified three heterozygous deletions encompassing the genes *OTX2* and *SOX2*. **(A)** Normalised log<sub>2</sub> ratios indicating two deletions of *OTX2* in CaseID 3000 and CaseID 3346. **(B)** Normalised log<sub>2</sub> ratios showing a deletion of *SOX2* in CaseID 2850.



**Supp. Figure S2.** Intragenic mutations identified in 12 families. A pedigree, mutation summary and sequence chromatogram(s) are shown for each family with an identified intragenic mutation: eight *SOX2* mutation families, three *OTX2* mutation families and one *STRA6* mutation family. Excepting the parents in the recessive *STRA6* family, all individuals carrying a mutation are indicated by CaseID. For each pedigree all known affected individuals are shown, and all individuals shown were

tested for the identified mutation. Unfilled symbols containing N represent unaffected individual with wild type sequence; solid filled symbol, affected individual carrying mutation(s); dotted filled symbol, unaffected individual carrying mutation. All sequence chromatograms show the forward read, except for FamID 2867 where the reverse read is shown for clarity of data in a Gln (Q) repeat region. The wild type reference sequence used for each gene is as follows, *SOX2*, NT\_005612.15 GI:88966845; *OTX2*, NT\_026437.11 GI:51493278; *STRA6*, NG\_009207.1 GI:219275560.