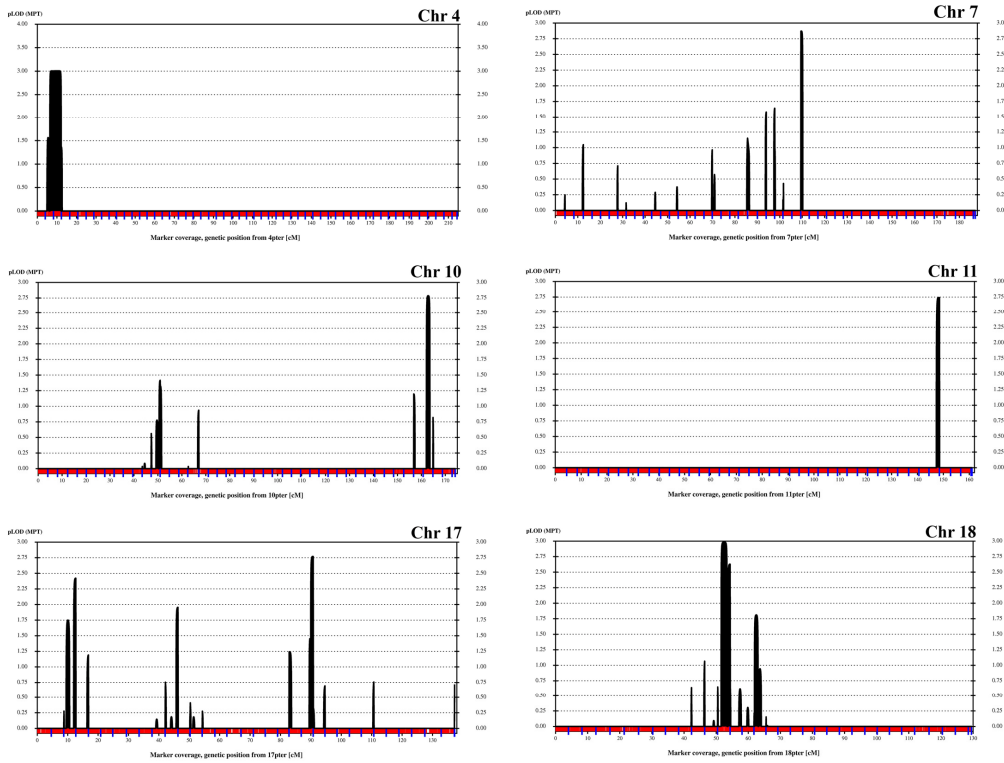
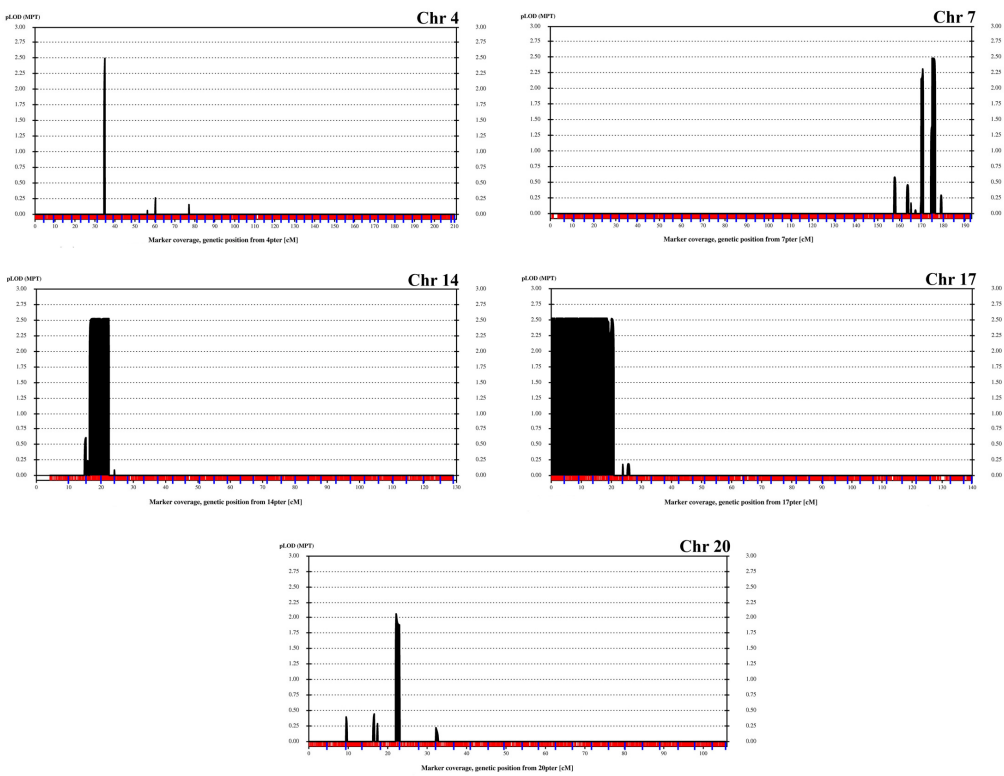


Supplementary Figure S1 Multipoint LOD scores based on a recessive model with full penetrance. Only chromosomes that yield scores >2 are presented.

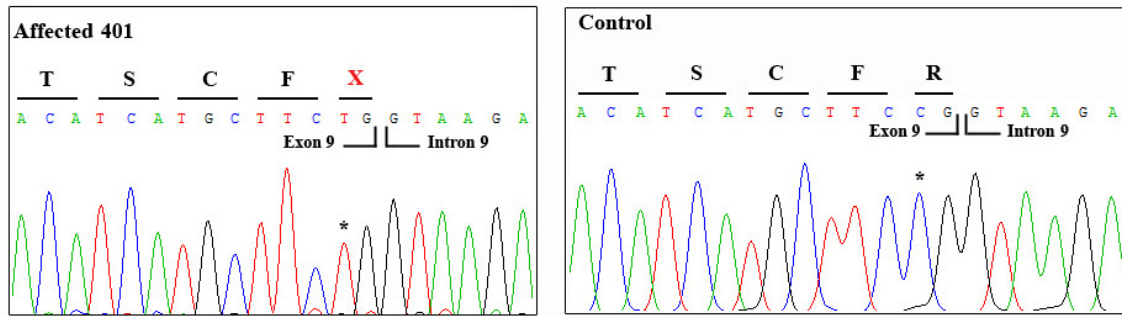
Family 1



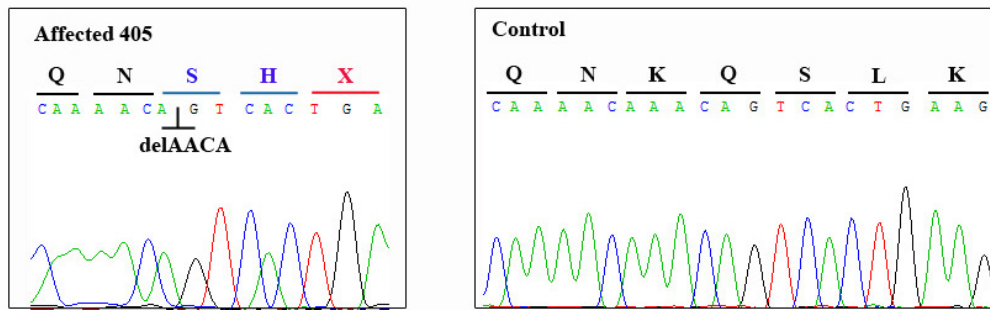
Family 2



Supplementary Figure S2 Chromatograms showing the identified mutations in a homozygous individual and the reference sequence.



TAF4B c.1831 C>T (p.R611X) in family 1



ZMYND15 c.1520delAACA (p.K507Sfs*3) in family 2

Supplementary Figure S3 Quantification of transcript levels for ZMYND15 isoforms 1 and 2 relative to POLR2A in various tissues.

