



Fig. S1. Sequence chromatograms showing nucleotide variants c.2T>C (cases 1 and 3) and c.3G>A (case 2) (black arrows) resulting in the p.Met1? mutation in the first codon (boxed) of KLHL24. The mutation is detected in the gDNA of the 3 probands and it is absent in the gDNA of their parents.