



**SUPPLEMENTARY FIG. S3.** Electropherograms (left panel) showing heterozygosity for a single nucleotide substitution (T/C) in P2 or wild-type sequence (T) in his parents. The right panel indicates heterozygosity for this nucleotide change in DNA from peripheral blood mononuclear (top), buccal mucosal (middle), and hair follicle (bottom) cells.