



SUPPLEMENTARY FIG. S2. Electropherograms (left panel) showing heterozygosity for a single nucleotide substitution (C/T) in P1 or wild-type sequence (C) 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.