**Suplementary Figure 1**. DHPLC pattern and sequence of identified mutations. The left side of each panel represents the DHPLC pattern of key individuals in families 1 (a), 2 (b), 3 (c), 4 (d), 5 (e), 6 (f), 7 (g), 8 (h). On the right side of each panel the mutated sequence is reported with the mutation written above. Note that the control (C) shows always a unique peak (homoduplex peak) while patients (P) show additional peaks.