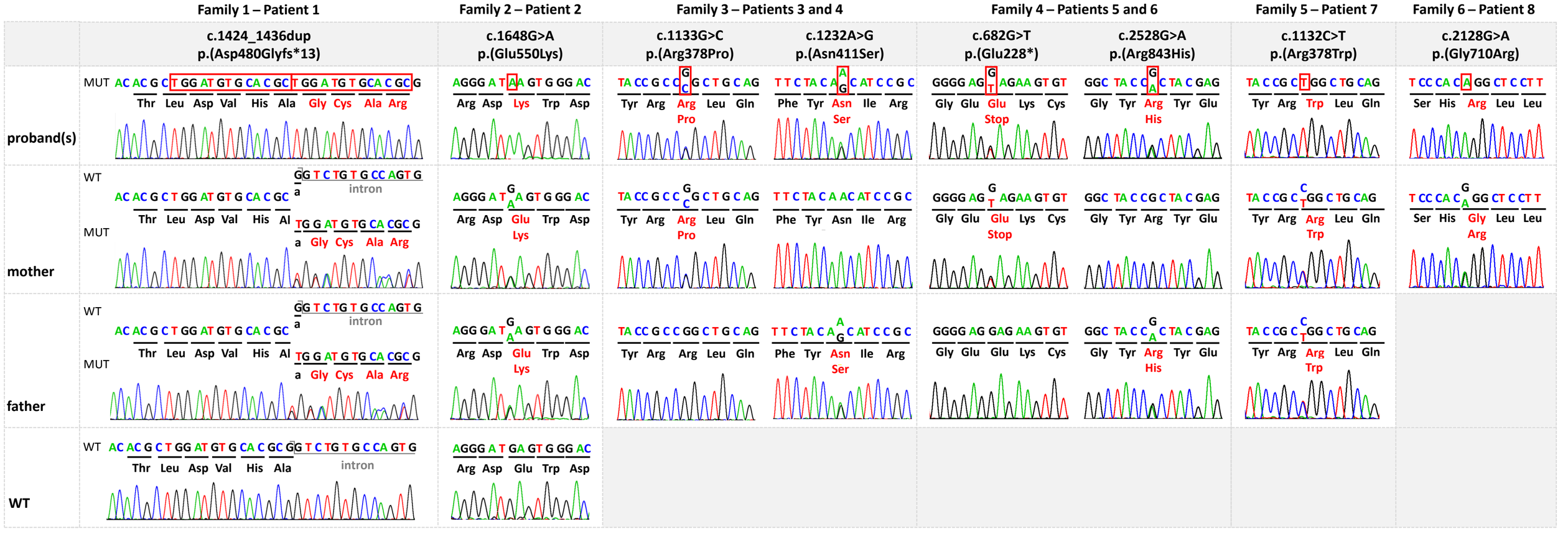
Supplementary Figure 1



Supplementary Figure 1. Identification or validation and segregation analysis of the identified *AMPD2* variants in the six families by Sanger-sequencing. Family and patient numbers as well as *AMPD2* variant description on DNA and protein level are given in the upper panel. Partial sequence electropherograms show the homozygous variant in patients 1, 2, 7, and 8 (families 1, 2, 5, and 6) as well as the compound heterozygous variants detected in patients 3 to 6 (families 3 and 4) (upper panel). The respective parts of the electropherograms of the patients' parents, who are heterozygous for the variant (families 1, 2, 5, and 6) or heterozygous for one of the two variants (families 3 and 4) are also shown. Nucleotide triplets and encoded amino acids (three-letter code) of the wild-type (WT) and mutated (MUT) allele are indicated above the electropherograms. The respective wild-type sequence is shown for families 1 and 2 in the bottom row. Mutated base(s) are highlighted by red boxes; changes in the AMPD2 amino acid sequence are indicated by red colored residues.