

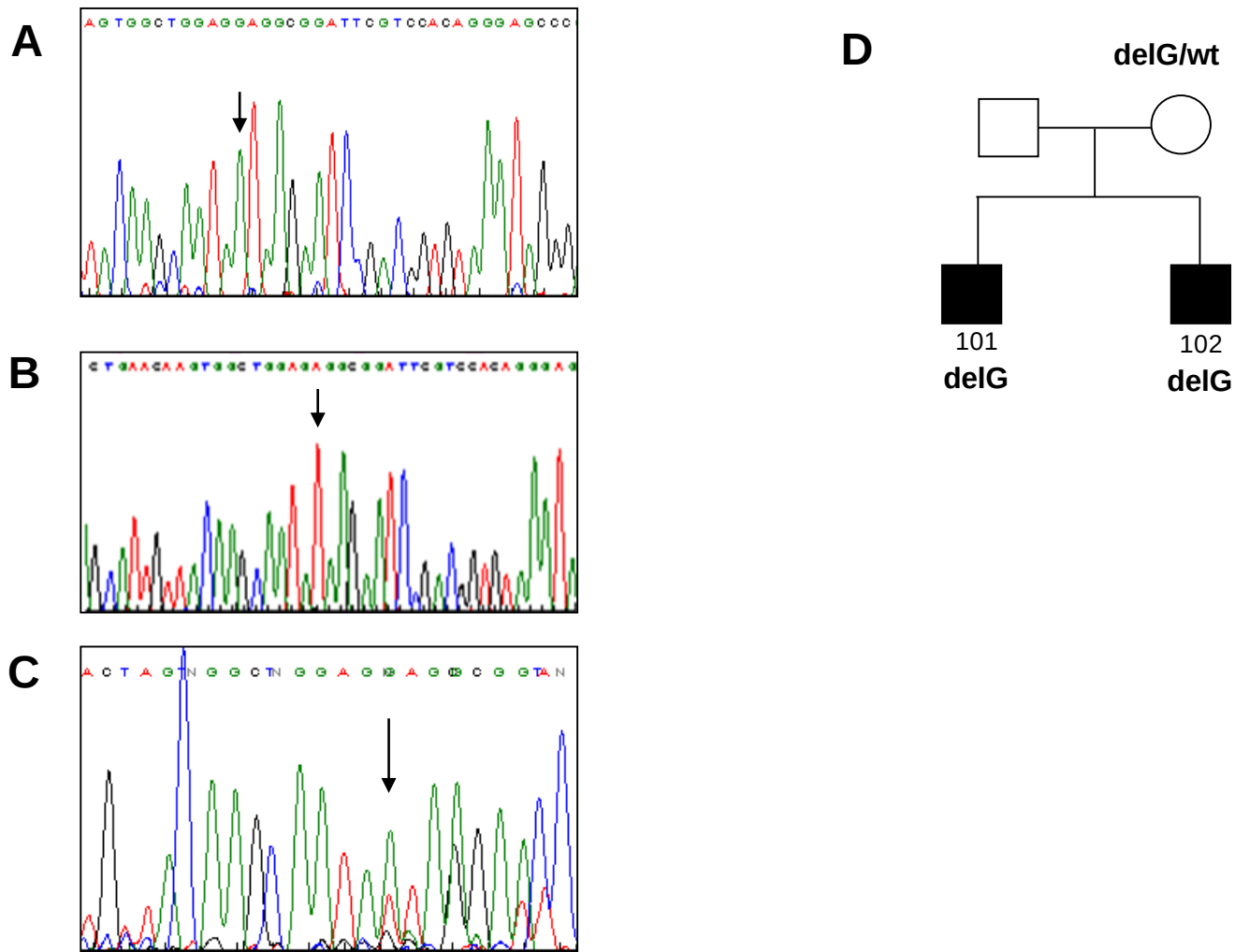


Figure S1: Data of the Family 295. Chromatograms showing wild type (1A), hemizygous NM_000307.4:c.772delG (p.(Glu258ArgfsX30)) mutation in the affected male (1B) and c.772delG (p.(Glu258ArgfsX30)) mutation in the female carrier (1C). The pedigree contains two affected male siblings (1D).

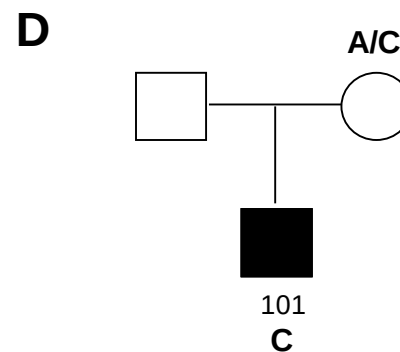
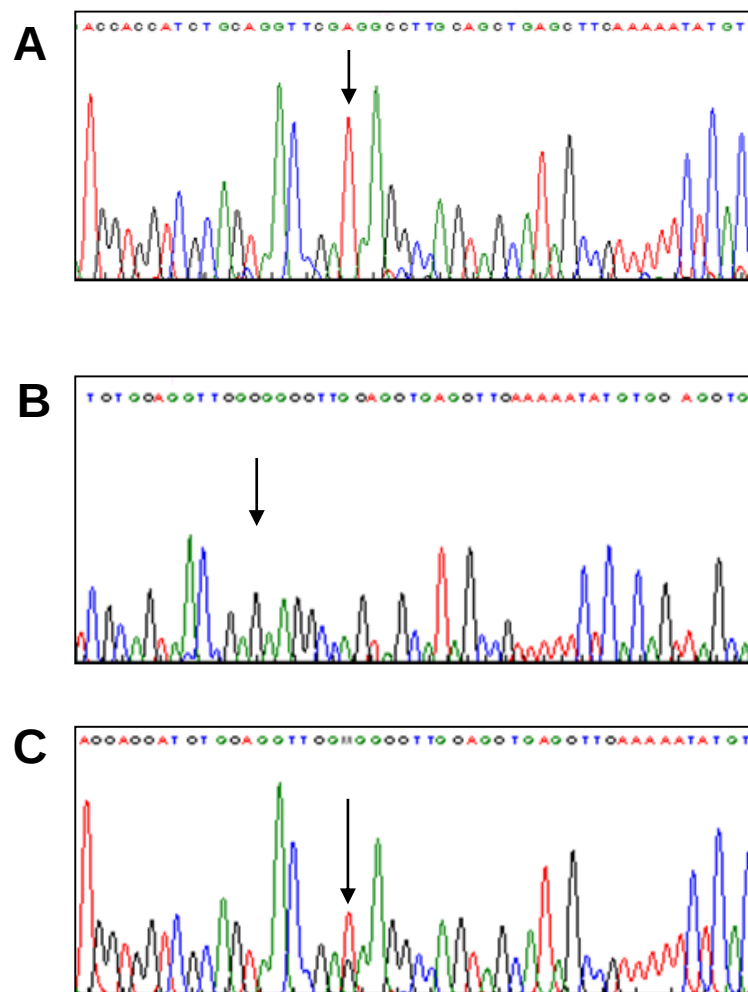


Figure S2: Data of the family 667. Chromatograms showing wild type (2A), hemizygous NM_000307.4:c.707A>C (p.(Glu236Ala)) mutation in the affected male (2B) and c.707A>C (p.(Glu236Ala)) mutation in the female carrier (2C). One affected male was in the family (2D).

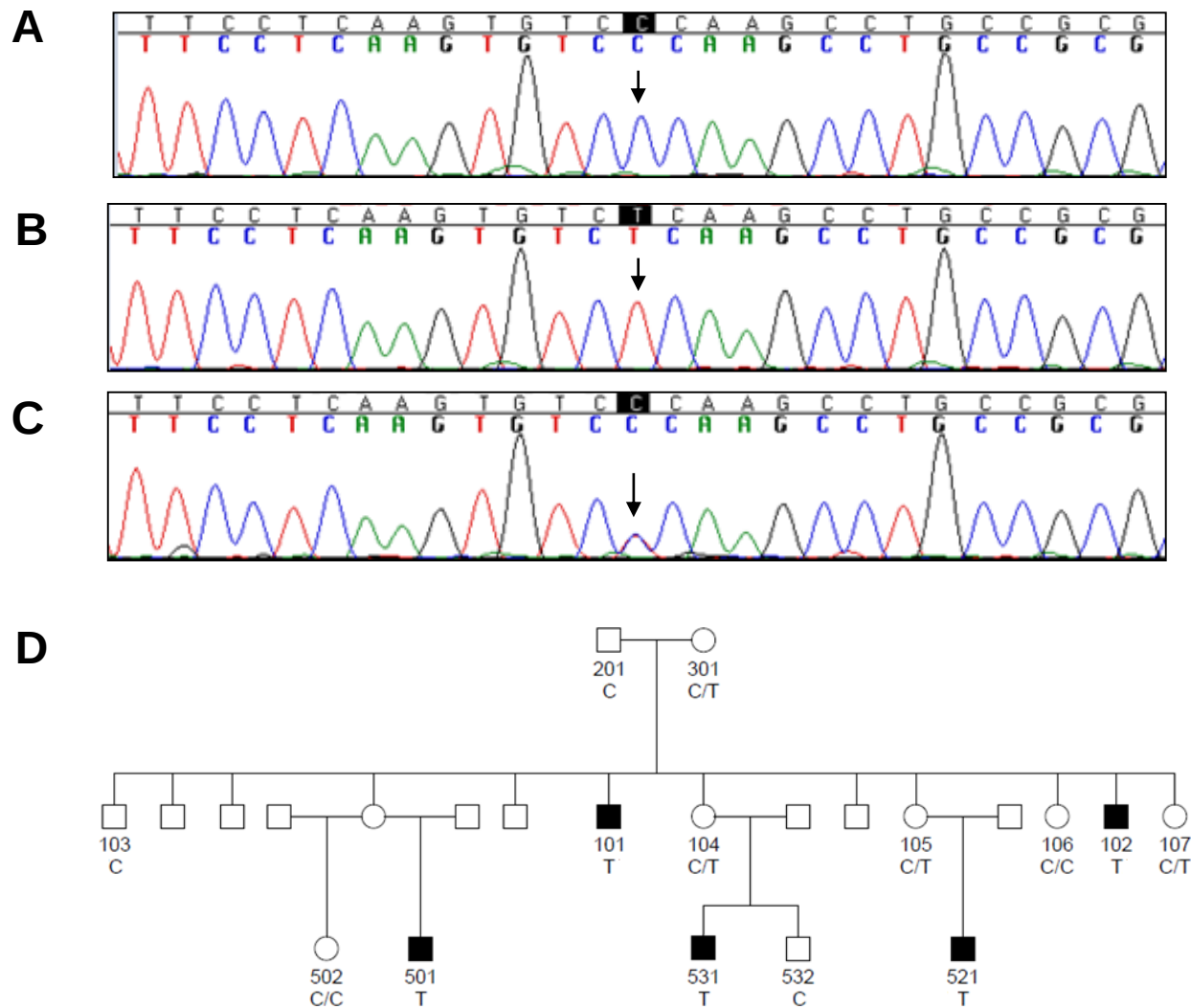
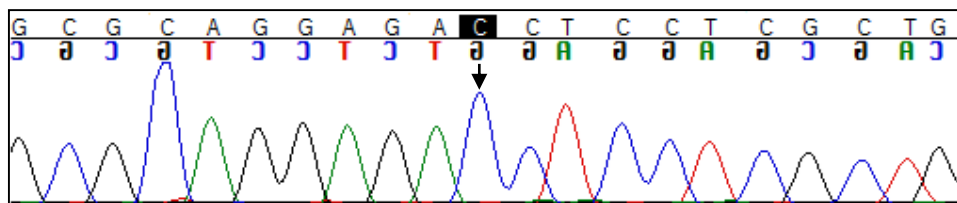
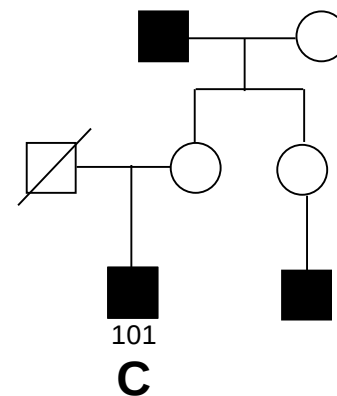


Figure S4: Data of the family 1225. Chromatograms showing wild type (4A), hemizygous NM_000307.4:c.902C>T (p.(Pro301Leu)) mutation in the affected male (4B) and c.902C>T (p.(Pro301Leu)) mutation in the female carrier (4C). The pedigree contains five affected males and is consistent with X-linked recessive inheritance (4D).

A**B**

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C

Figure S5: Data of the family 1535. Chromatogram showing hemizygous NM_000307.4:c.987T>C (p.(Ile308Thr)) mutation in the affected male (5A) There are three affected males and the pedigree is consistent with X-Linked recessive inheritance (5B).