Sanger sequencing is no longer always necessary based on a single-center validation of 1,109 NGS variants in 825 clinical exomes

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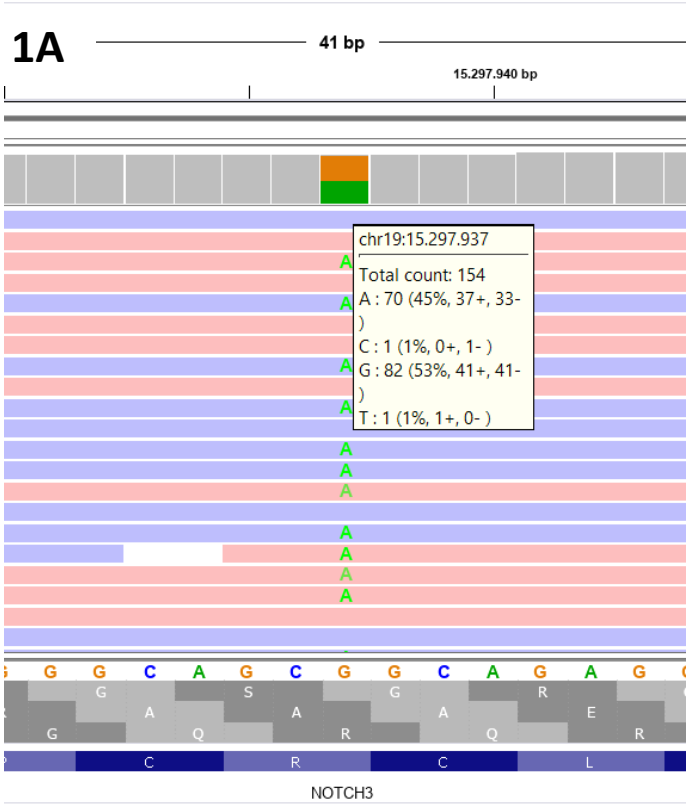
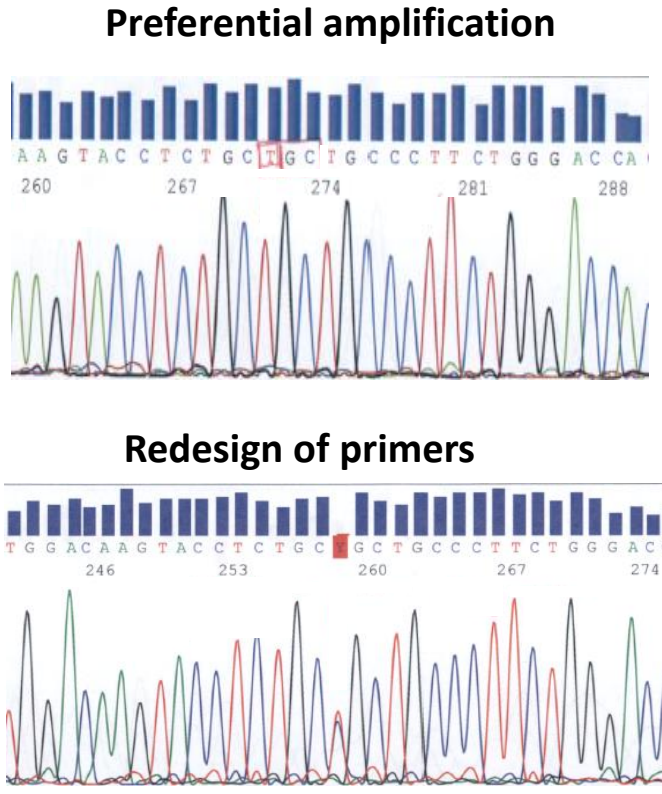
Supplementary Figure S1

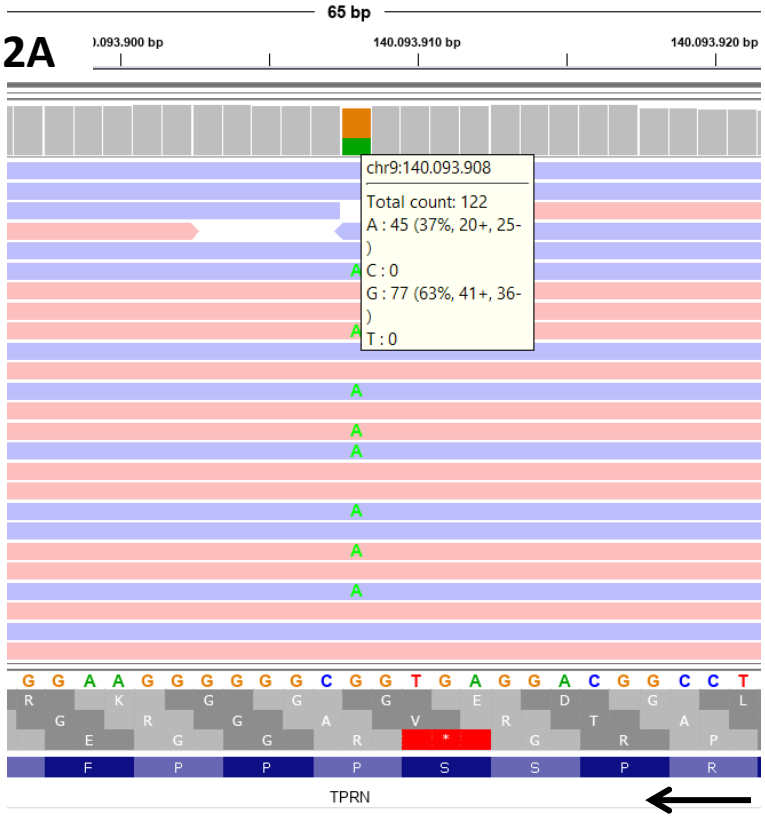
1A) IGV visualization of the heterozygous variant c.1819C>T (p.Arg607Cys) in the *NOTCH3* gene (NM_000435.2). 1B) Sanger chromatogram: the heterozygous variant is not detected. 1C) Sanger chromatogram: the heterozygous variant is only detected in the second round of amplification, after the redesign of primers. IGV=Integrative Genomics Viewer

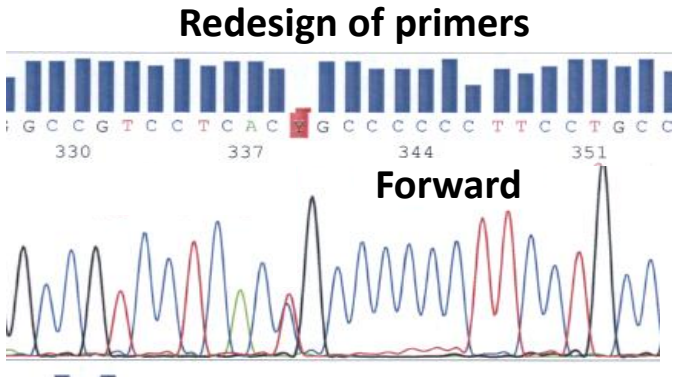


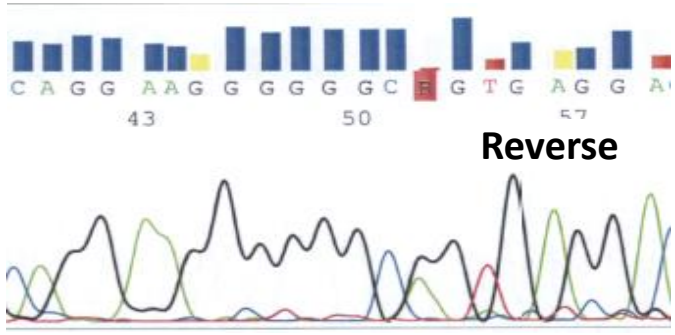


Supplementary Figure S2

2A) IGV visualization of the heterozygous variant c.1256C>T (p.Pro419Leu) in the *TPRN* gene (NM_001128228). 2) Sanger chromatogram: The variant is detected with both Forward (2B) and Reverse (2C), only after a redesign of primers. The sanger chromatogram showing the preferential amplification during the first round of PCR is not available. IGV=Integrative Genomics Viewer







Supplementary Figure S3

3A) IGV visualization of the heterozygous variant c.489C>G (p.Ser263Arg) in the *C1QTNF5* gene (NM_015645.4). 3B) Sanger chromatogram of the variant in the Peripheral blood and Buccal cells (in bold), showing a preferential amplification of the mutant allele in the affected patient but not in the peripheral blood of the affected cousin. IGV=Integrative Genomics Viewer