



Figure 7. Sanger sequencing chromatogram for patient 12008422, showing a heterozygous deletion removing two exons from the reading frame from the *USH2A* gene (chr1:216,167,537-216,177,486; *USH2A* c.6326-3582_6658-1028del, p.(Asp2109Glyfs*11), NM_206933.2). Heterozygous whole exon deletions have previously been reported as pathogenic in the *USH2A* gene.^{1,2}

1. Baux D, Blanchet C, Hamel C, et al. Enrichment of LOVD-USHbases with 152 *USH2A* Genotypes Defines an Extensive Mutational Spectrum and Highlights Missense Hotspots. *Hum Mutat* 2014.
2. Krawitz PM, Schiska D, Krüger U, et al. Screening for single nucleotide variants, small indels and exon deletions with a next-generation sequencing based gene panel approach for Usher syndrome. *Molecular Genetics & Genomic Medicine* 2014:n/a-n/a.