



**Figure 4. Sanger sequencing chromatogram for patient 12002355**, showing a heterozygous deletion removing the start codon and first 6 coding exons of the *PCDH15* gene (chr10:56,094,632-56,613,219; *PCDH15* c.-189197\_c.610-5166, NM\_001142763.1). Large structural rearrangements have previously been assigned as the cause of Usher syndrome in the *PCDH15* gene, including single and multiple exon deletions.<sup>1-3</sup>

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3. Glockle N, Kohl S, Mohr J, et al. Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. *Eur J Hum Genet* 2014;22:99-104.