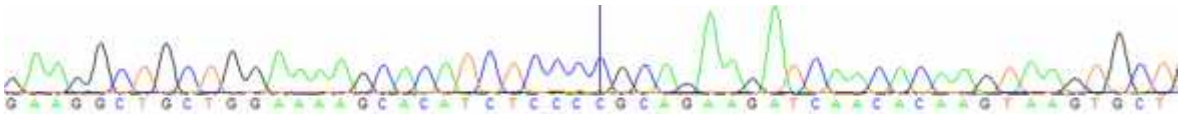

Figure 17. Sanger sequencing chromatogram for patient 11012959, showing a homozygous missense variant in the *TRPM1* gene (chr15:31,358,296; *TRPM1* c.707T>C, p.(Leu236Pro), NM_002420.5). c.707T>C has not previously been reported in patients with recessively inherited congenital stationary night blindness (CSNB), nor has it been reported as a normal polymorphism. However, missense mutations in *TRPM1* have previously been reported in a recessive state in patients with CSNB,[1-3] and *in-silico* analysis suggests that c.707T>C will have a detrimental effect on the TRPM1 protein.

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2. Sergouniotis PI, Chakarova C, Murphy C, et al. Biallelic variants in TTLL5, encoding a tubulin glutamylase, cause retinal dystrophy. Am J Hum Genet 2014;94:760-9.
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