



**Figure 15. Sanger sequencing chromatogram for patient 065238**, showing a heterozygous intronic variant in the *ABCA4* gene (chr1:94,476,351; *ABCA4* c.5714+5G>A, NM\_000350.2). c.5714+5G>A reduces the splicing efficiency of the exon40/intron40 donor splice site<sup>1</sup> and has previously been reported as pathogenic.<sup>2,3</sup>

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