



Figure S1: Variant phasing of *ROM1* variants using PacBio long-read amplicon sequencing. Primers were designed to amplify 1.2 Kb across *ROM1* variants c.339dupG and c.712del in proband 079830 and long-read amplicon sequencing was performed using PacBio. HiFi reads were visualized in IGV and grouped by phase to reveal that both variants are in *trans*.