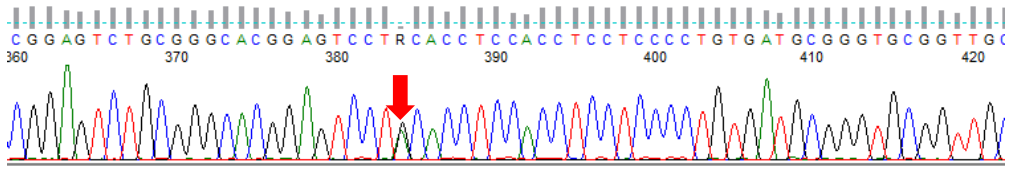
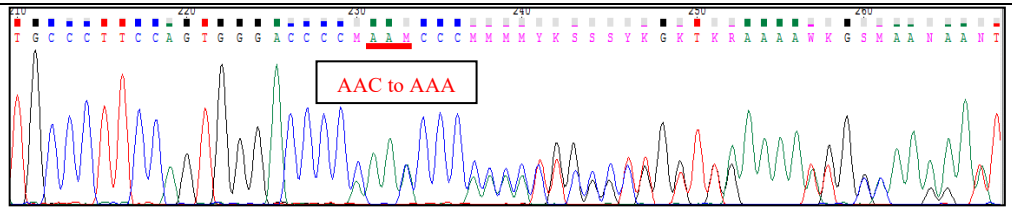
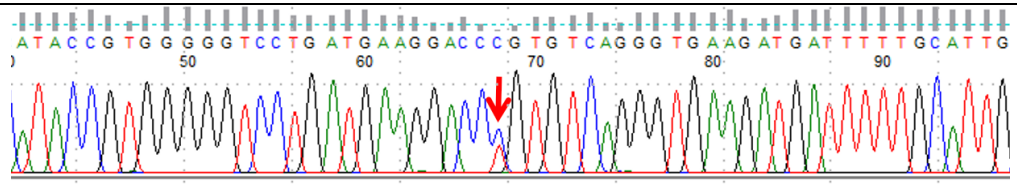
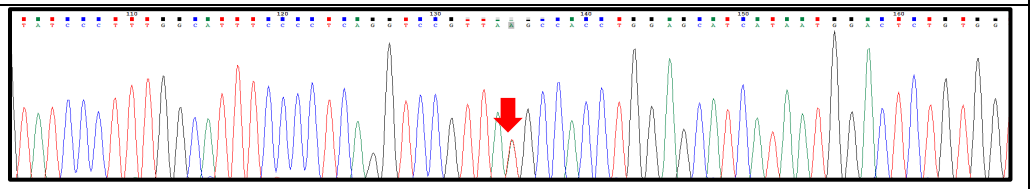
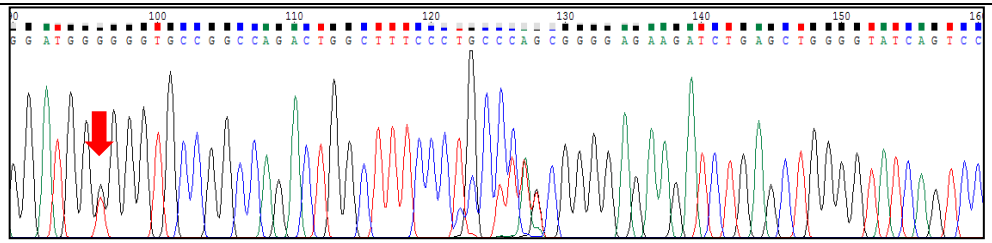
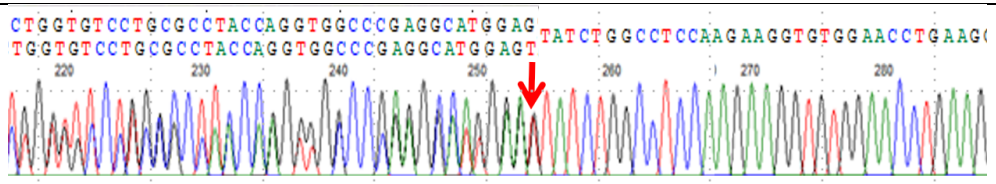
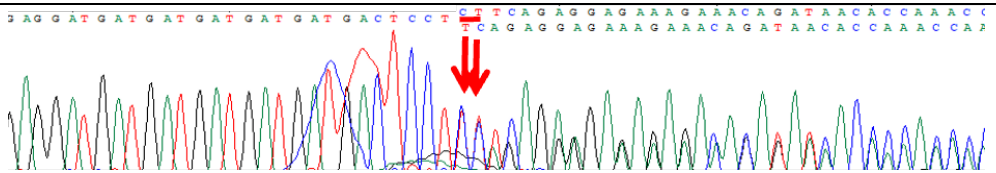
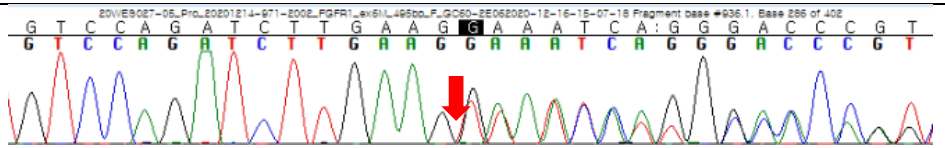
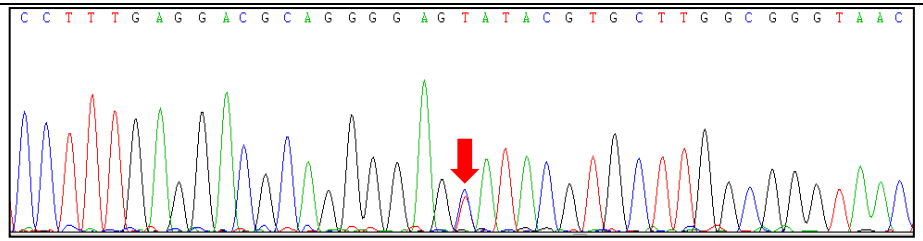
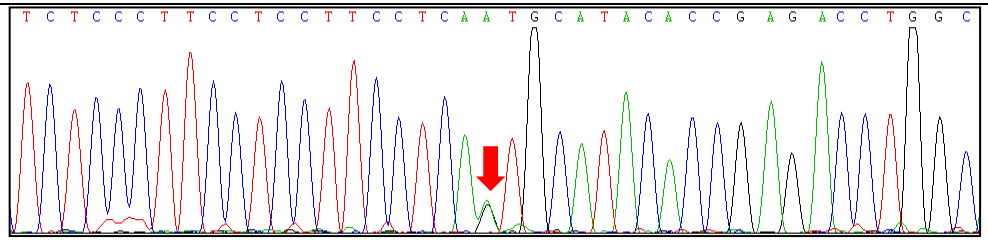
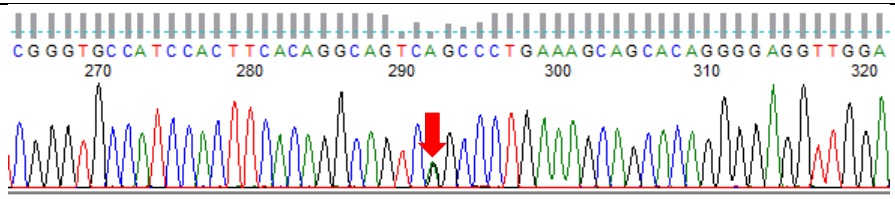
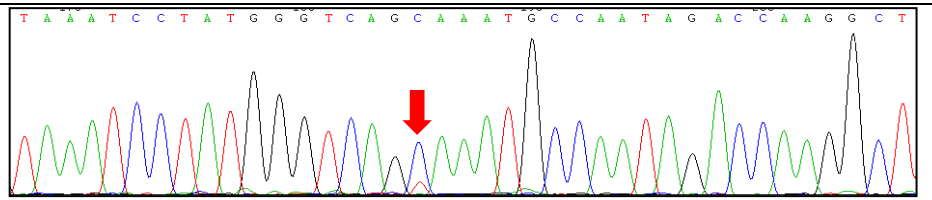
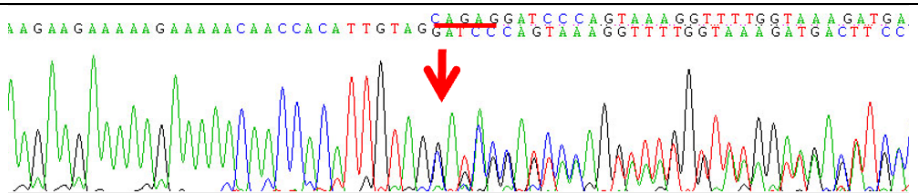
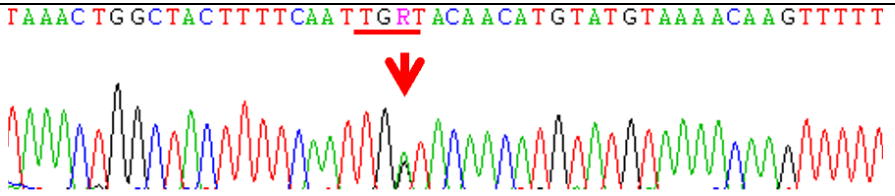
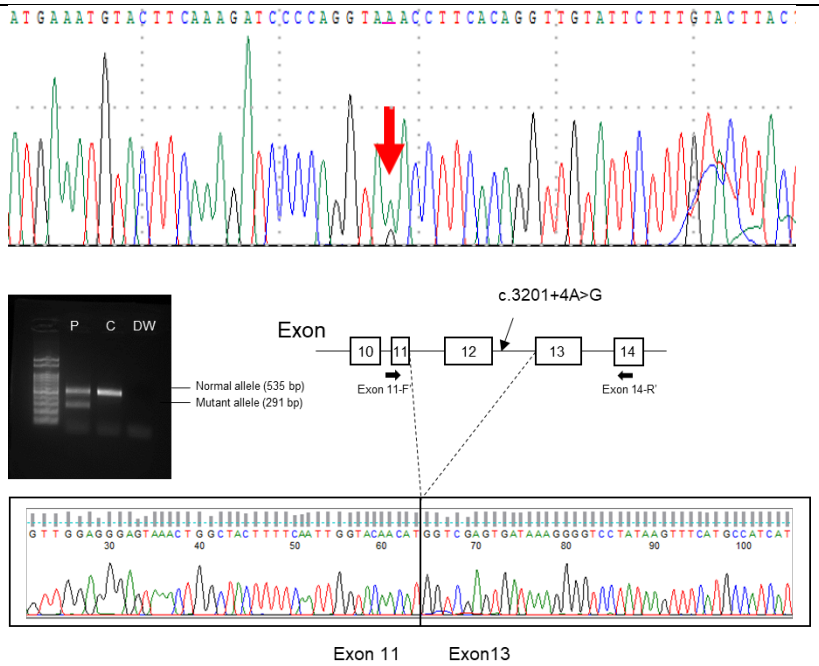
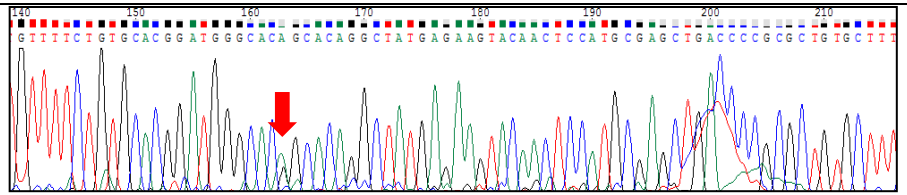
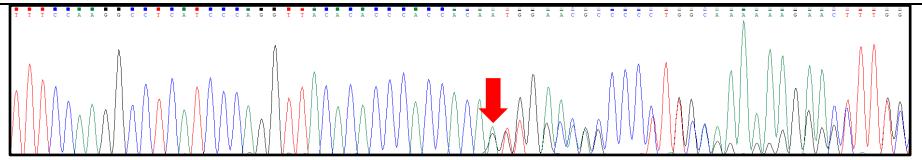
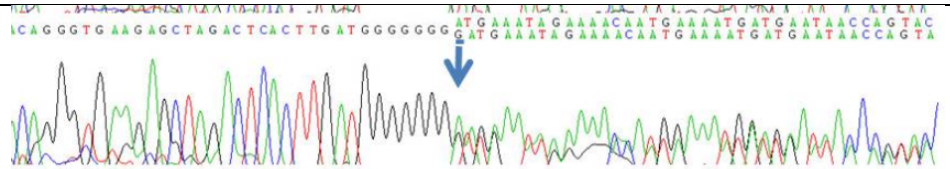
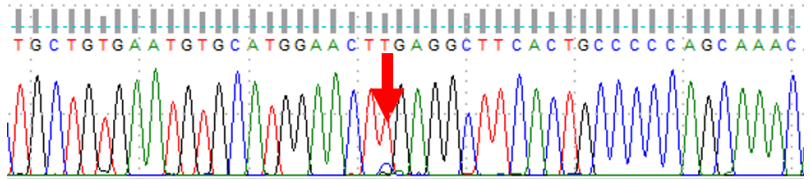
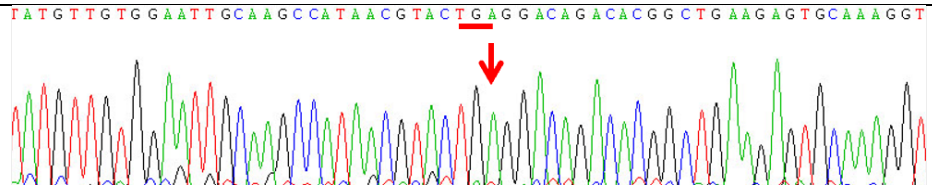
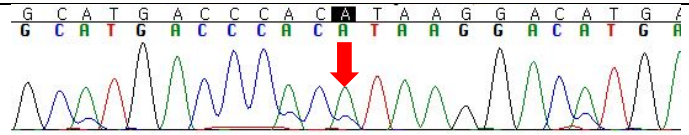
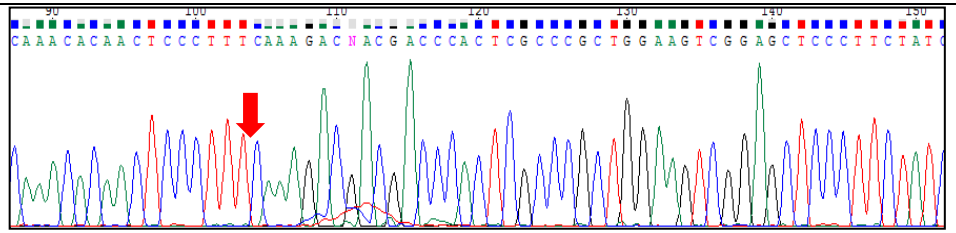
Supplementary Table 3. Sanger sequencing confirmation of the rare genetic variants identified by targeted gene panel sequencing, whole-exome sequencing, and whole-genome sequencing.

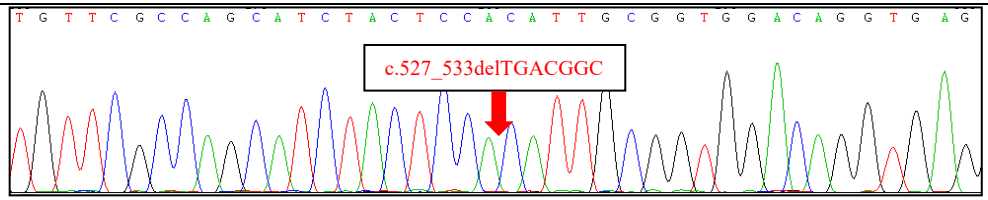
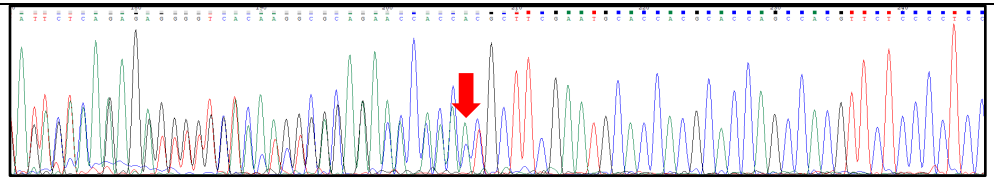
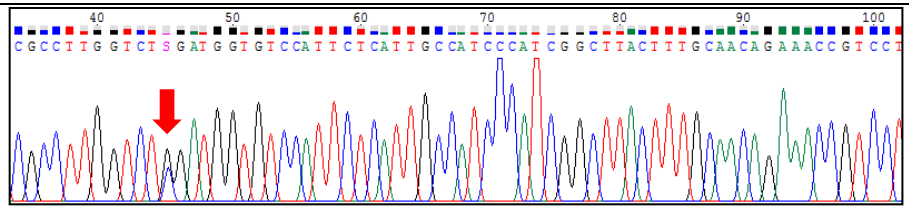
Gene	Nucleotide change	Amino acid change	ACMG/AMP criteria	Electropherogram
FGFR1	c.265C>T	p.Q89*	Likely pathogenic	
FGFR1	c.551dup	p.N185Kfs*16	Likely pathogenic	
GNRHR	c.719G>A	p.R240Q	Likely pathogenic	
FGFR1	c.630T>A	p.Y210*	Likely pathogenic	

FGFR1	c.1663+2T>G	Splice site	Likely pathogenic	
FGFR1	c.1837dup	p.Y613Lfs*43	Likely pathogenic	
FGFR1	c.404_405del	p.S135Ffs*8	Likely pathogenic	
FGFR1	c.936+1del	Splice site	Likely pathogenic	
FGFR1	c.1015T>C	p. Y339H	Likely pathogenic	

FGFR1	c.1855-1G>A	Splice site	Pathogenic	
FGFR1	c.1981C>T	p.R661*	Pathogenic	
CHD7	c.118C>T	p.Q40*	Likely pathogenic	
CHD7	c.1823_1827del	p.A608Gfs*4	Likely pathogenic	
CHD7	c.2949G>A	p.W983*	Likely pathogenic	

CHD7	c.3201+4A>G	Splice site	Likely pathogenic (skipping of exon 12)	
CHD7	c.5405-7G>A	Splice site	Likely pathogenic	
CHD7	c.7059dup	p.V2354Sfs*12	Likely pathogenic	

CHD7	c.8962dup	p.D2988Gfs*2	Likely pathogenic	
ANOS1	c.814C>T	p.R272*	Pathogenic	
ANOS1	c.1140G>A	p.W380*	Likely pathogenic	
ANOS1	c.1449+1G>A	Splice site	Likely pathogenic	
ANOS1	c.1260del	p.Q421Kfs*61	Likely pathogenic	

TACR3	c.527_533del	p.M176Tfs*14	Pathogenic	
GNRH1	c.190_191insCT	p.R64Pfs*17	Likely pathogenic	
PROKR2	c.533G>C	p.W178S	Likely pathogenic	

Sequence variants confirmed by Sanger sequencing are indicated by red arrows.