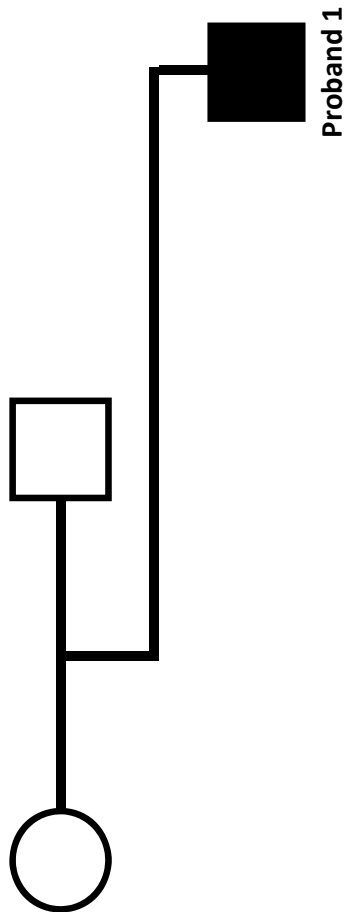


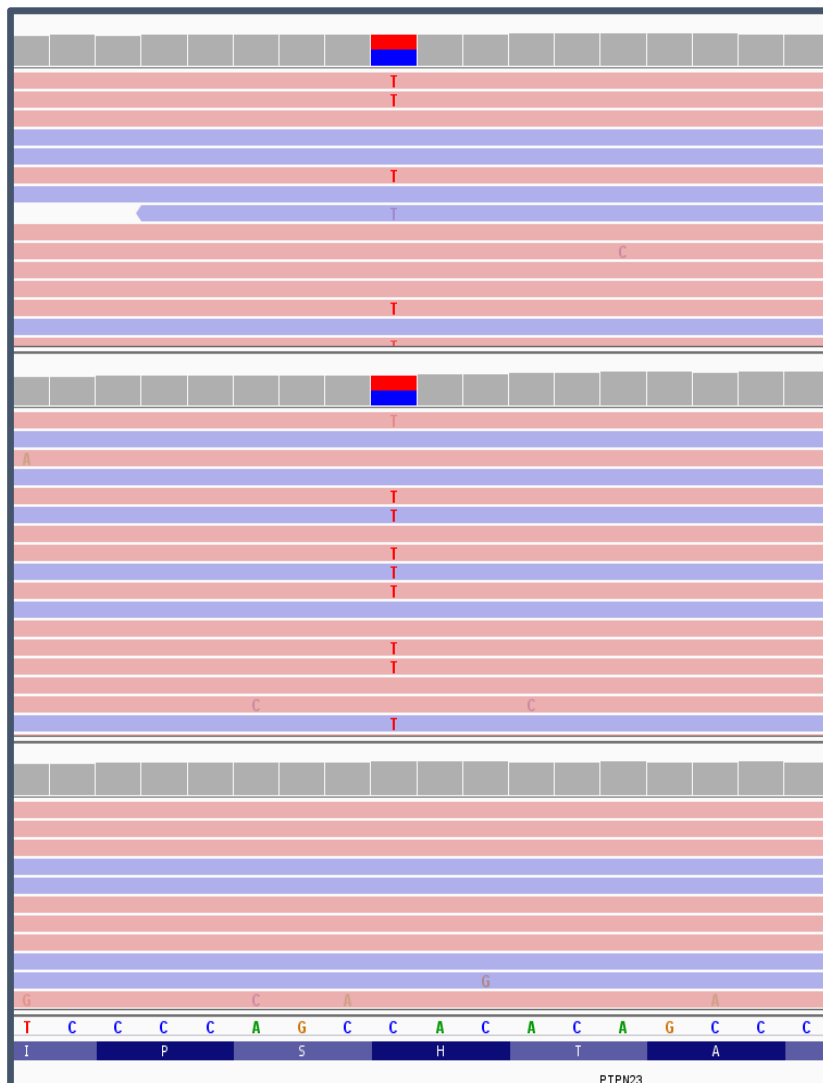
Phenotype and mutation spectrum expansion of the *PTPN23* associated disorder characterized by neurodevelopmental delay and structural brain abnormalities

Renee Bend, Lior Cohen, Melissa T. Carter, Michael J. Lyons, Dmitriy Niyazov, Mohamad A. Mikati, Samantha K. Rojas, Richard E. Person, Yue Si, Ingrid M. Wentzensen, Regeneron Genetics Center, Erin Torti, Jennifer A. Lee, Kym M. Boycott, Lina Basel-Salmon, Claudia Gonzaga-Jauregui, Carlos R. Ferreira

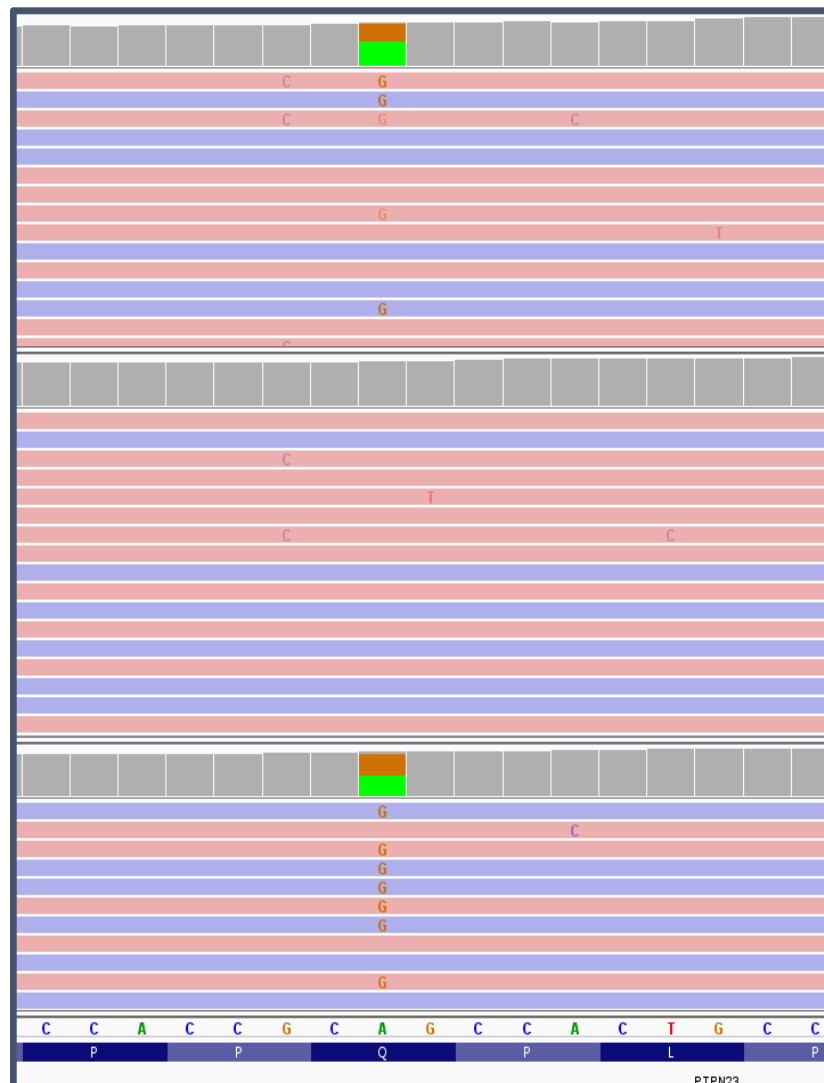
Supplementary Material

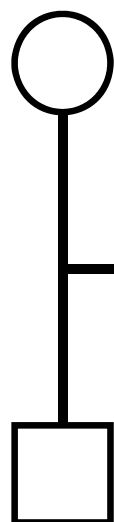


chr3:47410478(C>T)
c.2680C>T; p.His894Tyr

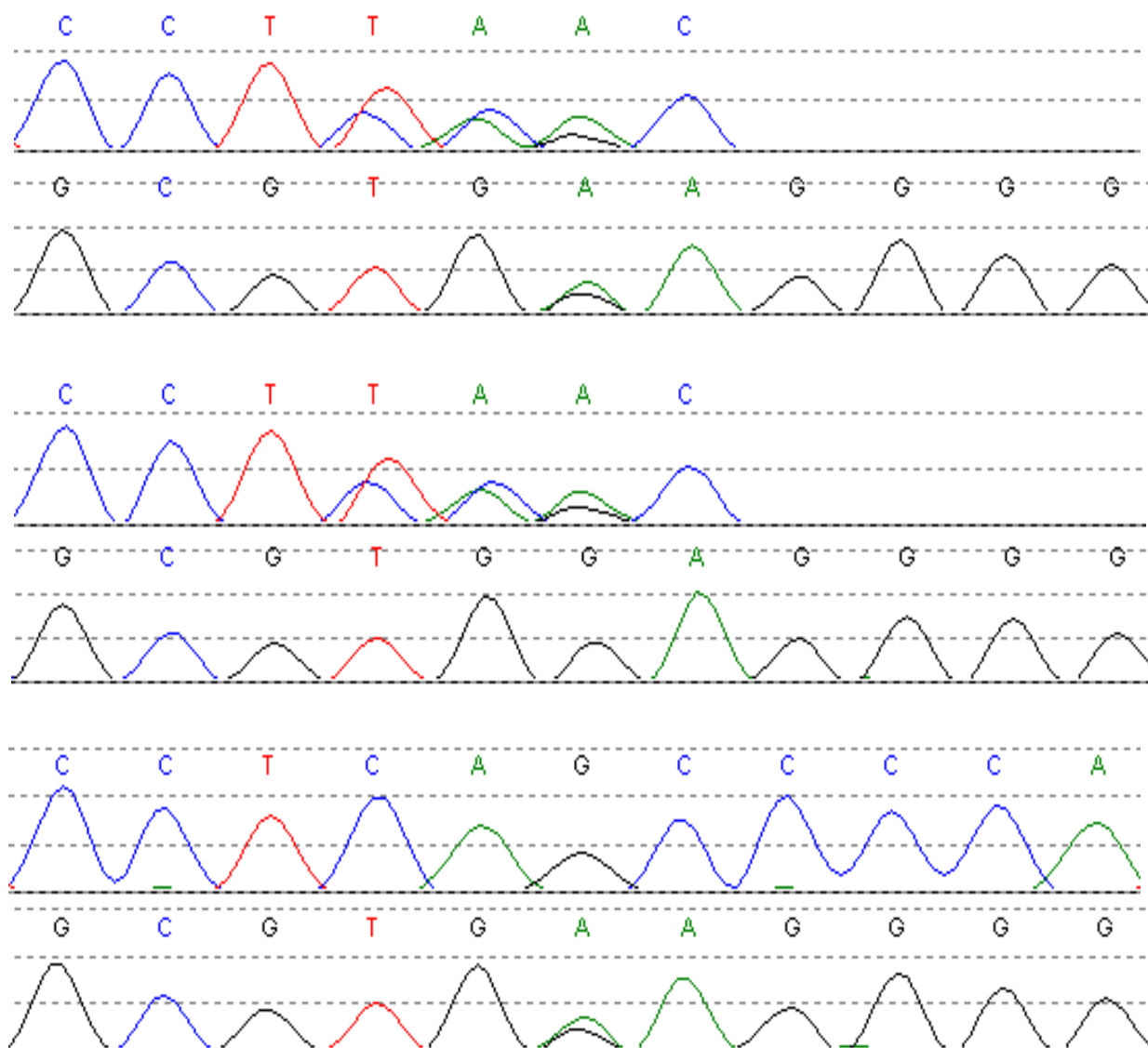


chr3:47410545(A>G)
c.2747A>G; p.Gln916Arg





Proband 2



chr3:47410676-47410688
(delCAGCCCCATCCT);
c.2878_2889del12;
p.Gln960_Pro963del

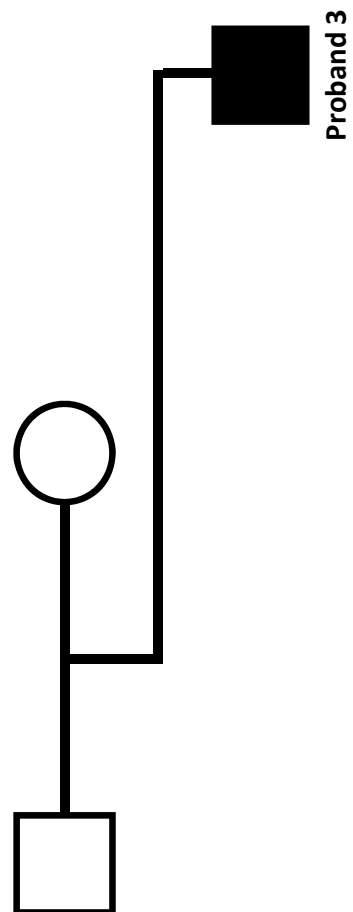
chr3:47411546(G>A)
c.3748G>A; p.Glu1250Lys

chr3:47410676-47410688
(delCAGCCCCATCCT);
c.2878_2889del12;
p.Gln960_Pro963del

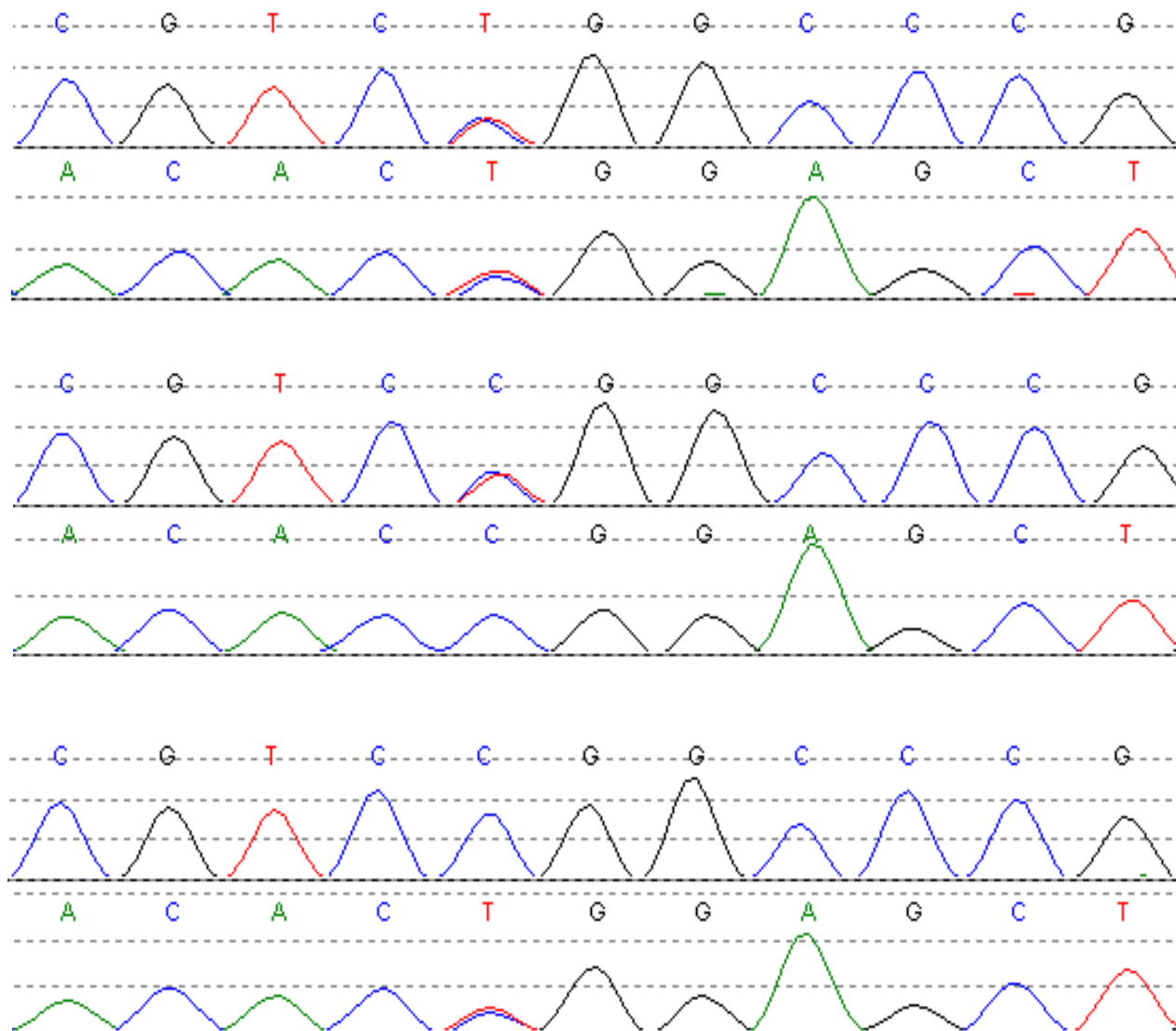
chr3:47411546(G/G)

chr3:47410676-47410688 (WT)

chr3:47411546(G>A)
c.3748G>A; p.Glu1250Lys



Proband 3



chr3:47408451(C>T)
c.1291C>T; p.Arg431Trp

3:47410284(C>T)
c.2486C>T; p.Pro829Leu

chr3:47408451(C>T)
c.1291C>T; p.Arg431Trp

3:47410284(C/C)

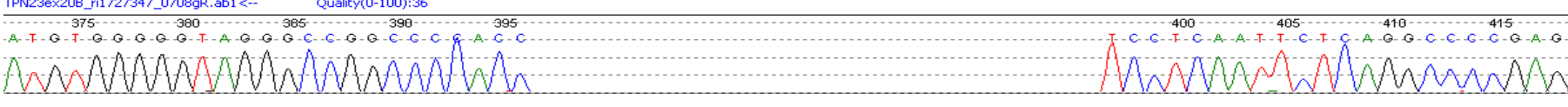
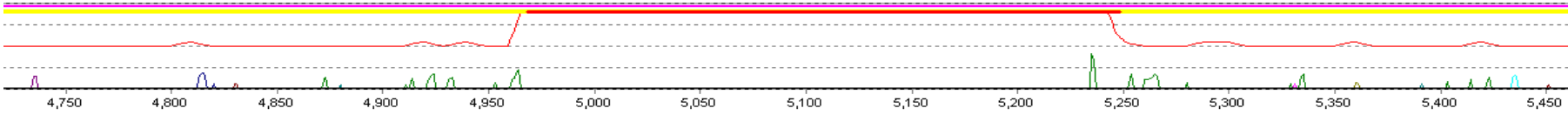
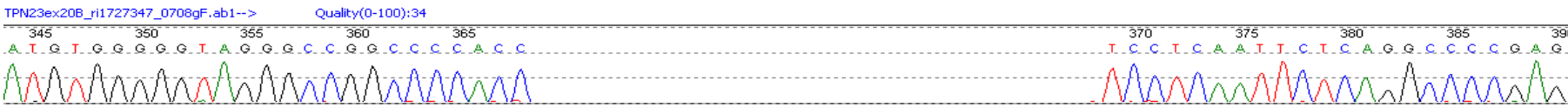
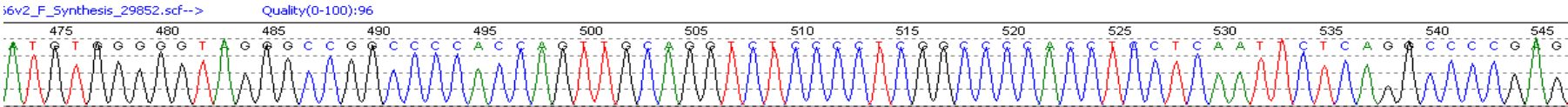
chr3:47408451(C/C)

3:47410284(C>T)
c.2486C>T; p.Pro829Leu

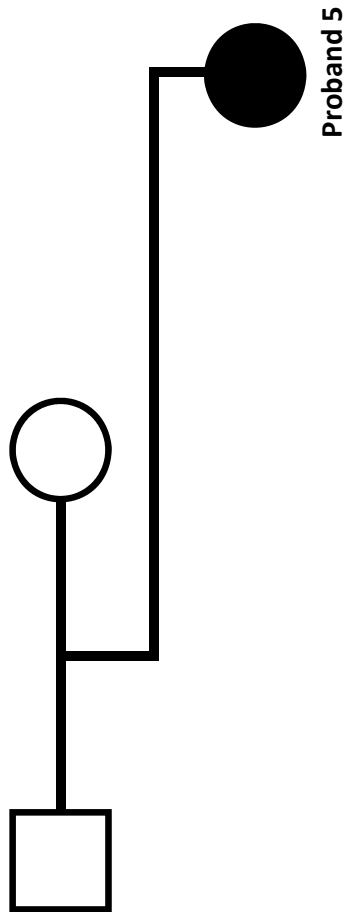
chr3:47410366-47410392 (delAGTTGCAGGTCTCCCCTCGGCCCCACC); c.2568_2594del27; p.Val857_Pro865del

29950	29955	29960	29965	29970	29975	29980	29985	29990	29995	30000	30005	30010	30015	30020
A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G	A T G T G G G G G T A G G G C C C C C A G T T G C A G G T C T C C C T C G G C C C A C C T C C C T C A A T T T C T C A G G C C C G A G

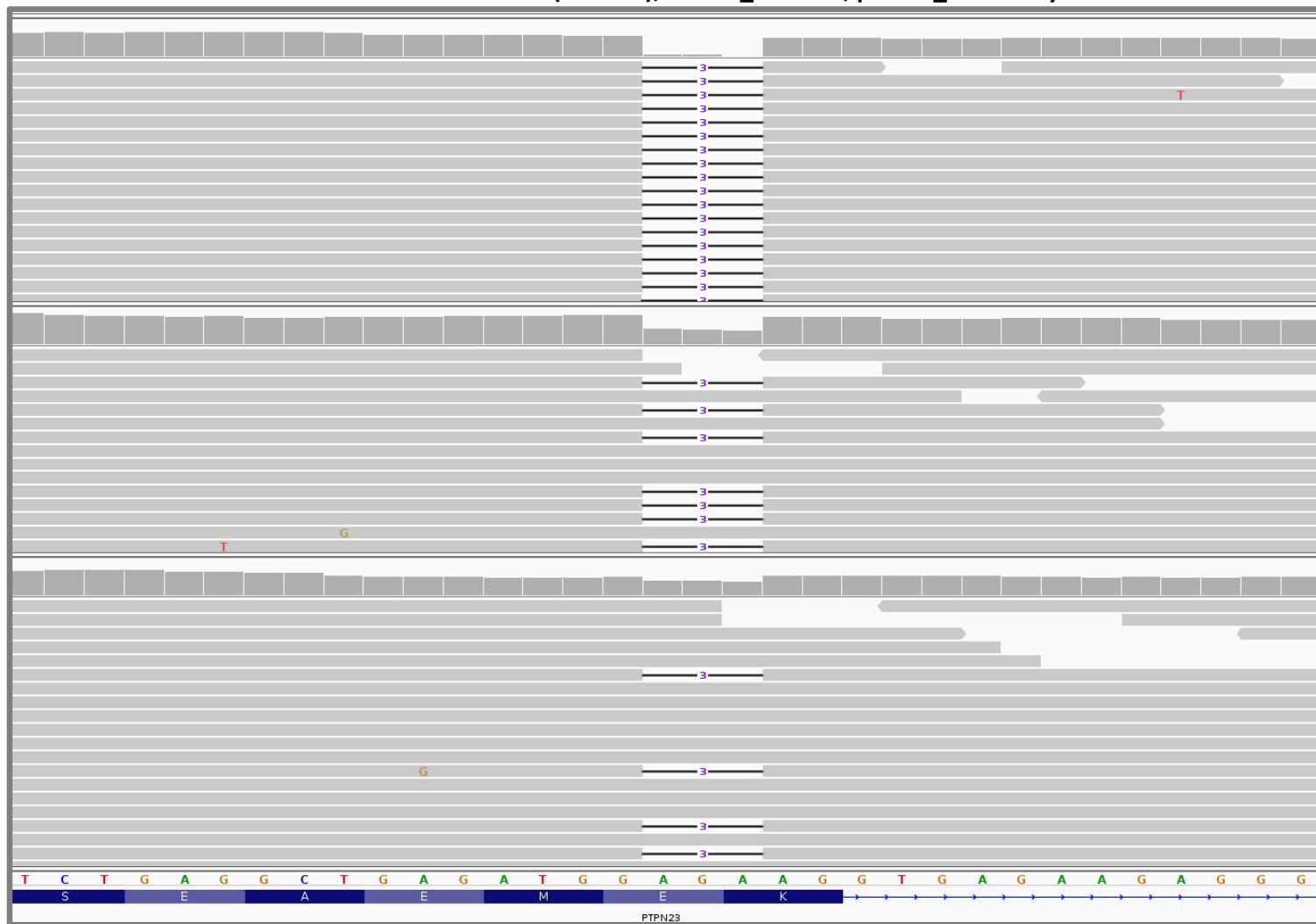
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Y	V	G	V	G	P	A	P	P	V	A	G	L	P	S	A	P	P	P	Q	F	S	G	P	E
						c.2560						c.2580							c.2600					

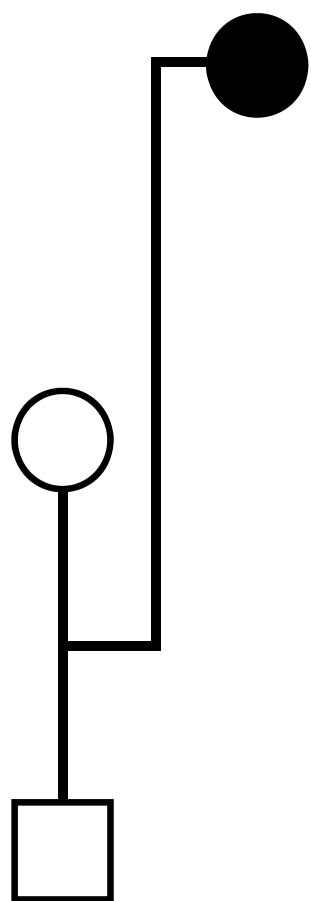


Proband 4

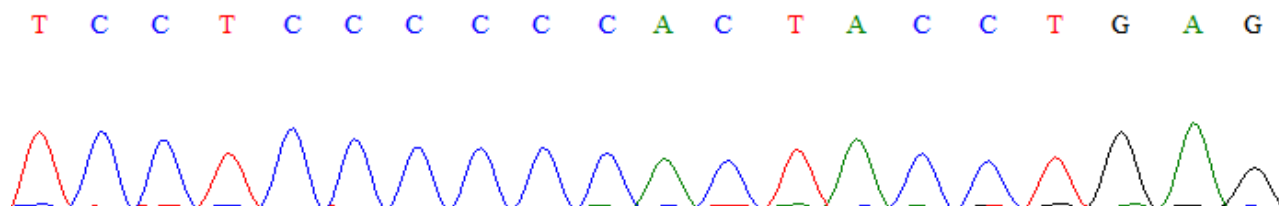


chr3:47411682-47411684(delAGA); c.3884_3886del; p.1295_1296delLys

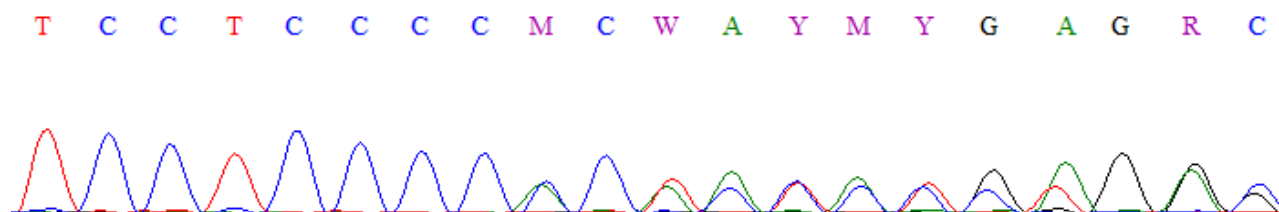




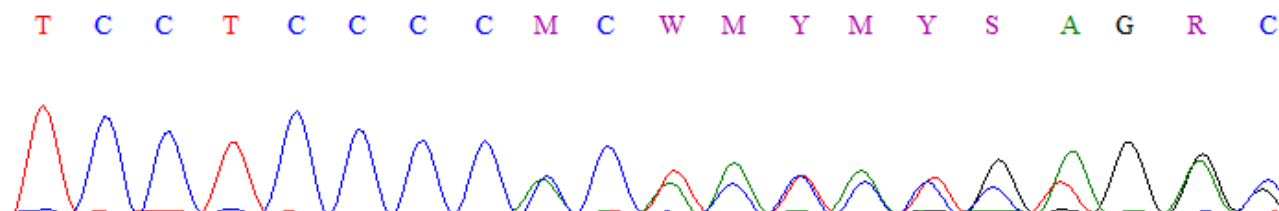
Proband 6



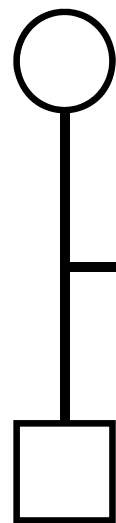
chr3:47412926(insCC);
c.4651_4652dup;
p.Leu1552Hisfs*33
[homozygous]



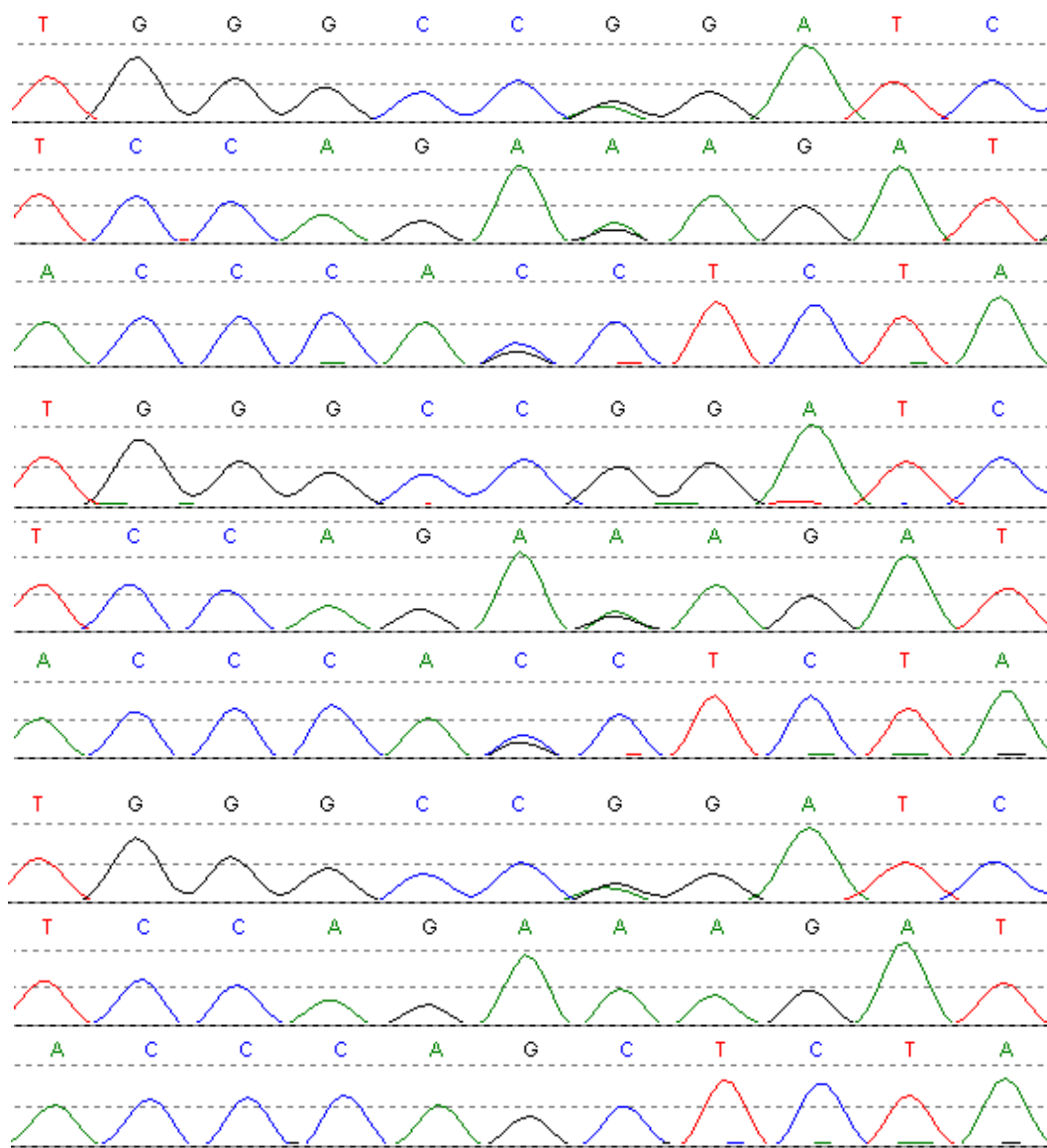
chr3:47412926(insCC);
c.4651_4652dup;
p.Leu1552Hisfs*33
[heterozygous]



chr3:47412926(insCC);
c.4651_4652dup;
p.Leu1552Hisfs*33
[heterozygous]



Proband 7



chr3:47406548(G>A); c.695G>A; p.Arg232Gln
chr3:47409268(A>G); c.1748A>G; p.Lys583Arg
chr3:47410849(G>C); c.3051G>C; p.Gln1017His
chr3:47406548(G/G)
chr3:47409268(A>G); c.1748A>G; p.Lys583Arg
chr3:47410849(G>C); c.3051G>C; p.Gln1017His
chr3:47406548(G>A); c.695G>A; p.Arg232Gln
chr3:47409268(A/A)
chr3:47410849(G/G)