1 Snapshot

This patient, [redacted] was analysed to identify genetic determinants for the clinical diagnosis of phenotype [redacted] (and phenotype subgroup [redacted] . The age [redacted] of onset was [redacted]. The following candidate causal variant(s) were identified with the use of `Design DNA SNV INDEL v1` pipeline, which implements `Design QV SNV INDEL v1` criteria, with the controlled dataset `Design DNA SNV INDEL v1 release`. This pipeline is concerned with WGS germline short variant discovery (SNVs + Indels) and interpretation.

Guru score range: ≤ -7, -1 to -6, 0 to 5, 6 to 9, ≥10

Impact range: MODIFIER, LOW, MODERATE, HIGH

Patient candidate variant 1

Patient ID:	[redacted]
Guru score:	31
Impact:	MODERATE
GRCh38:	chr[redacted]C/G
Gene symbol:	[redacted]
Consequence:	missense variant
HGVSc (cDNA change):	ENST00000[redacted]: c.[redacted]>C
HGVSp (Protein change):	ENSP00000[redacted]: p[redacted]Ser
Genotype:	Homozygous
gnomAD allele frequency:	NA
Compound heterozygous flag:	1
ACMG count:	4
Patient max score:	31
ACMG criteria:	PS1, PS5, PM3, PP3