Variant classification according to the NGSnPPGL recommendations	
Pathogenic	Variant reported in literature with strong evidence of pathogenicity or Null variant with functional evidence for pathogenicity
Likely Pathogenic	Null variant with no material available for functional study or Missense variant with ≥3 *in silico* predictions in favour of pathogenicity and functional study supportive of a damaging effect or Intronic or silent variant with predicted splice impact by *in silico* analysis and and functional study supportive of a damaging effect
VUS	Insufficient evidence to classify or Contradictory criteria
Likely Benign	Missense variant with ≥3 in silico predictions in favour of the variant being benign or Intronic or silent variant with no predicted splice impact or Co-occurrence with pathogenic variant or Functional evidence for non-pathogenicity
Benign	AF>1% in control groups or Presence in control groups with no co-segregation with the disease or AF=0,01-1% and functional evidence for non-pathogenicity

Supplemental Figure 2: Variant classification according to the NGSnPPGL recommendations