Single Nucleotide Variant	Minor Allele Frequency	Reference (DOI)
Ala.391.Thr	• 0.05 in American populations • 0.08 in Northern European populations • 0.14-0.25 in Ashkamzi Jewish populations • Monomorphic in African and South Asian populations	10.1172/jci.insight.140978
Gly.38.Arg	• 0.0001255 in European populations	10.1016/j.ajhg.2015.11.003
Ile.340.Asn Val.33.met Ser.335.Thr Gly.204.Cys Cys.113.Ser	• Unknown frequency • Identified using penetrant autosomal recessive models with a rare disease allele frequency of 0.0001	10.1007/s10545-016-0010-6 10.1016/j.ajhg.2015.11.002 10.1016/j.ajhg.2015.11.003