

Phenotype Consistency	Points per proband				
	Heterozygous Probands (PS4)	Homozygous/Compound Heterozygous Probands (PM3)			
		VBC is confirmed in trans with a P/LP variant	VBC is assumed in trans with a P/LP variant	VBC is confirmed in trans with a VUS	VBC is homozygous
Phenotype specific with gene (meets PP4) and all relevant genes for disorder tested (SCID gene panel or exome/genome sequencing), with no other variant of interest	+1 point	+2 point	+1.5 point	+1.0 point	+1.0 point
Phenotype specific with gene (meets PP4) but not all relevant genes for disorder tested, and/or more than one variant of interest	+0.5 point	+1 point	+0.75 point	+0.5 point	+0.5 point
Phenotype consistent with gene (has at least one core feature, see PP4) and all relevant genes for disorder tested (SCID gene panel or exome/genome sequencing), with no other variant of interest	+0.5 point	+0.5 point	+0.5 point	+0.5 point	+0.5 point
Phenotype consistent with gene (has at least one core feature, see PP4) but not all relevant genes for disorder tested, and/or more than one variant of interest	+0.25 point	+0.25 point	+0.25 point	+0.25 point	+0.25 point
Phenotype not consistent with gene	+0 point	+0 point	+0 point	+0 point	+0 point

*VBC variant being curated