

Points system for assigning weight of evidence for PM3
ClinGen Lysosomal Diseases Variant Curation Expert Panel

Date: June 7, 2024

Based on ClinGen Sequence Variant Interpretation Working Group guidance-
https://clinicalgenome.org/site/assets/files/3717/svi_proposal_for_pm3_criterion_-_version_1.pdf

Classification/Zygosity of other variant	Points per Proband	
	Confirmed in trans	Phase unknown
<i>Pathogenic or Likely pathogenic variant</i>	1.0	0.5 (P) 0.25 (LP)
<i>Homozygous occurrence (max points = 1.0)</i>	0.5	N/A

Note: The VCEP does not give points for compound heterozygotes if the second variant is a VUS.

PM3 Point Table			
PM3_Supporting	PM3	PM3_Strong	PM3_VeryStrong
0.5 points	1.0 points	2.0 points	4.0 points