PS2 *De Novo* Table

Table 1. Points* awarded per *de novo* occurrence

Phenotypic consistency	Points per Proband	
	***de novo* with confirmed parental relationships**	***de novo* with unconfirmed parental relationships**
Phenotype highly specific for gene	2	1
Phenotype consistent with gene but not highly specific	1	0.5

*Note that these points are *not* equivalent to the points used to classify a variant per the Tavtigian et al 2020 "Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines"

Table 2. Recommendation for determining the appropriate ACMG/AMP evidence strength level for *de novo* occurrence(s)

Supporting (PS2_Supporting or PM6_Supporting)	**Moderate** (PS2_Moderate or PM6)	**Strong** (PS2 or PM6_Strong)	**Very Strong** (PS2_VeryStrong or PM6_VeryStrong)
0.5	**1**	**2**	**4**

Source: https://clinicalgenome.org/site/assets/files/3461/svi_proposal_for_de_novo_criteria_v1_1.pdf