

Table 4a:

Recommendations for PP1 (segregation evidence)

	General Recommendations		
	Supporting	Moderate	Strong
Likelihood	4:1	16:1	32:1
LOD Score	0.6	1.2	1.5
Autosomal dominant threshold	2 affected segregations	4 affected segregations	5 affected segregations
Autosomal recessive threshold	See Table 4b	See Table 4b	See Table4b

Table 4b:

Recommendations for autosomal recessive segregation evidence (PP1)

		General Recommendations (Phenocopy not an issue)											
		Unaffected Recessive Segregations											
		0	1	2	3	4	5	6	7	8	9	10	
Affected segregations	0	0	0.12	0.25	0.37	0.5	0.62	0.75	0.87	1	1.12	1.25	
	1	0.6	0.73	0.85	0.98	1.1	1.23	1.35	1.48	1.6	1.73	1.85	
	2	1.2	1.33	1.45	1.58	1.7	1.83	1.95	2.08	2.2	2.33	2.45	
	3	1.81	1.93	2.06	2.18	2.31	2.43	2.56	2.68	2.81	2.93	3.06	
	4	2.41	2.53	2.66	2.78	2.91	3.03	3.16	3.28	3.41	3.53	3.66	
	5	3.01	3.14	3.26	3.39	3.51	3.63	3.76	3.88	4.01	4.13	4.26	
	6	3.61	3.74	3.86	3.99	4.11	4.24	4.36	4.49	4.61	4.74	4.86	
	7	4.21	4.34	4.46	4.59	4.71	4.84	4.96	5.09	5.21	5.34	5.46	
	8	4.82	4.94	5.07	5.19	5.32	5.44	5.57	5.69	5.82	5.94	6.07	
	9	5.42	5.54	5.67	5.79	5.92	6.04	6.17	6.29	6.42	6.54	6.67	
	10	6.02	6.15	6.27	6.4	6.52	6.65	6.77	6.9	7.02	7.15	7.27	

Affected segregations are counted in rows and unaffected segregations in columns. Affected segregations are affected family members in whom biallelic compound heterozygous or homozygous variants segregates. Unaffected segregations are defined as unaffected family members, typically siblings, who are at risk to inherit the two variants identified in the proband. These individuals should be either wild-type for both variants identified in the proband, or a heterozygous carrier for a single variant. Unaffected, carrier parents DO NOT count as unaffected segregations. There may be scenarios where individuals other than siblings could be counted as segregations, such as in families where one parent is affected with the autosomal recessive disorder, in large families with multiple branches, or in consanguineous families.

Each cell shows the LOD score of each combination of affected and unaffected segregations. LOD scores were calculated using a simplified LOD score formula, as described in Strande et al., 2017.