PS2/PM6 Codes

Note that **all probands** being considered for any pathogenic phenotype codes (e.g. PP1, PP4, PM6, PS2, PS4) at any strength must have the following phenotype characteristics:

- affected males should have some functional vision impairment by age 13
- with either foveo-macular changes or ERG measurement of a subnormal B wave

RS1 PS2/PM6 Table 1. Points awarded per *de novo* occurrence.

Points per de novo occurrence for X-linked IRD genes	*RS1* Points per Proband	
	Confirmed *de novo* with confirmed maternity	Assumed de novo with assumed maternity
Phenotype highly specific for gene – Requires OTC (optical coherence tomography) showing schisis	2	1
Phenotype consistent with gene but not highly specific – use when lacking OTC data.	1	0.5

Table 2. RS1 Recommendation for determining the appropriate ACMG/AMP evidence strength level for *de novo* occurrence(s)

Supporting	Moderate	Strong	Very Strong
0.5	1	2	4