**PS4:**

Notes:

- If variant meets criteria for BS1 or BA1, PS4 cannot apply
- Due to phenotypic variability (late onset, variable expressivity, incomplete penetrance) we cannot exclude the possibility of pathogenic variants being present in population databases.

**Proband counting: Please refer to PP4 document for diagnostic criteria**

| **Category** | **Total points required** *(cumulative points)* | **Individual proband points/ phenotype** *(points per proband)* |
|---|---|---|
| PS4_Very strong | 15+ | AxD definite diagnosis: +3 points<br>AxD probable diagnosis: +2 point<br>AxD consistent features: +1 points |
| PS4_Strong | 9-14 | AxD definite diagnosis: +3 points<br>AxD probable diagnosis: +2 point<br>AxD consistent features: +1 points |
| PS4_Moderate | 4-8 | AxD definite diagnosis: +3 points<br>AxD probable diagnosis: +2 point<br>AxD consistent features: +1 points |
| PS4_Supporting | 2-3 | AxD definite diagnosis: +3 points<br>AxD probable diagnosis: +2 point<br>AxD consistent features: +1 points |

**Citations**


- Yoshida et al 2017 PIMD: 23903069
- Prust et al 2011 PIMD: 21917775, Li 2005 PIMD:15732097
- Unpublished GLIA-CTN biorepository data
- Test variants from these following publications were used to test the point system:
    - Li 2005 (infantile, late onset) PIMD:15732097
    - Prust 2011 (185 reported) PIMD: 21917775