**Appendix 2: *RP1* allele frequencies and predicted pathogenicity**

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| --- | --- | --- | --- | --- | --- |
| **Variant** | **gnomAD****Frequency1** | **SIFT;****PolyPhen-2** | **MutationTaster** | **Classification2**  | **Patient(s)** |
| c.121T>Cp.(Tyr41His) | 14/282874 | Deleterious; Probably damaging | Polymorphism | Likely Pathogenic | CEI29345 |
| c.126G>Ap.(Lys42Asn) | 1/251482 | Deleterious; Probably damaging | Disease causing | Likely Pathogenic | MEE5 |
| c.139dupp.(Gln47Profs\*15) | 1/251486 | n/a | Disease causing | Likely Pathogenic | CEI24459 |
| c.312\_315delCCTAp.(Leu105Valfs\*10) | Absent | n/a | Disease causing | Likely Pathogenic | MEE5 |
| c.491C>Gp.(Pro164Arg) | 10/248874 | Deleterious; Probably damaging | Disease causing | Likely Pathogenic | MEE13 |
| c.515T>Gp.(Leu172Arg) | 24/248984 | Deleterious; Probably damaging | Polymorphism | Likely Pathogenic | CEI23745, CEI26528, CEI26529, CEI29023, CEI29345 |
| c.606C>Ap.(Asp202Glu) | Absent | Deleterious; Probably damaging | Polymorphism | Likely Pathogenic | MEE1, MEE2, MEE3, MEE4 |
| c.668delp.(Gly223Glufs\*41) | 8/282842 | n/a | Disease causing | Likely Pathogenic | MEE10, MEE11 |
| c.1126C>Tp.(Arg376\*) | 4/251268 | Deleterious; n/a | Disease causing | Pathogenic | MEE10 |
| c.1199\_1200delp.(Gln400Argfs\*18) | Absent | n/a | Disease causing | Likely Pathogenic | MEE13 |
| c.1234dupAp.(Met412Asnfs\*7) | 1/ 251326 | n/a | Disease causing | Pathogenic | MEE12 |
| c.1462delGp.(Glu488Lysfs\*44) | Absent | n/a | Disease causing | Pathogenic | MEE7 |
| c.1468G>Tp.(Glu490\*) | 7/251332 | Deleterious; n/a | Disease causing | Likely Pathogenic | MEE11 |
| c.1598\_1601delp.(Arg533Lysfs\*12) | 1/250640 | n/a | Disease causing | Pathogenic | CEI29023 |
| c.3155delTp.(Tyr1053Thrfs\*4) | 2/251118 | n/a | Disease causing | Pathogenic | CEI23745 |
| c.3428delAp.(Asn1143Ilefs\*25) | Absent | n/a | Disease causing | Pathogenic | MEE8 |
| c.4171delC p.(Gln1391Lysfs\*7) | Absent | n/a | Disease causing | Likely Pathogenic | MEE12 |
| c.4582\_4585delATCAp.(Ile1528Valfs\*10) | Absent | n/a | Disease causing | Pathogenic | CEI26528, CEI26529 |
| c.4788delTp.(Asp1597Thrfs\*29) | Absent | n/a | Disease causing | Likely Pathogenic | MEE9 |
| c.5017delCp.(Tyr1673Metfs\*37) | 2/251170 | n/a | Disease causing | Likely Pathogenic | CEI26396 |
| c.5428G>Tp.(Glu1750\*) | Absent | Deleterious; n/a | Disease causing | Pathogenic | CEI24459 |

1 gnomAD frequencies obtained 1/31/2010.

2 Classification based upon American College of Medical Genetics guidelines (*Genet Med*. 2015 May; 17(5): 405–424. doi:10.1038/gim.2015.30)