**Supplementary Table 1.** Variants in the human *AMH* gene published after the exhaustive review by Picard et al., 2017.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient** | **Origin** | **Exon-intron** | **Mutation** | **Effect** | **Alleles** | **Reference** | **Condition** |
| 1 | ? | 1 | NM\_000479.3:c.203\_203delC | NM\_000479.3:p.(p. L70Cfs\*7) | Homo | Mazen, 2017 | PMDS |
| 2 | ? | 1 | NM\_000479.3:c.283C>T | NM\_000479.3:p.(Arg95\*) | Hetero (compound) | Hughes, 2019 | PMDS |
| 2 | ? | 5 | NM\_000479.3:c.905G>A | NM\_000479.3:p.(Arg302Gln) | Hetero (compound) | Hughes, 2019 | PMDS |
| 3 | Turkey | 1 | NM\_000479.3:c.301G>A | NM\_000479.3:p.(Gly101Arg) | Homo | Bugrul, 2020 | PMDS |
| 4 | Turkey | 3 | NM\_000479.3:c.563G>A | NM\_000479.3:p.(Cys188Tyr) | Homo | Ata, 2021 | PMDS |
| 5 | ? | 3 | NM\_000479.3:c.649C>T | NM\_000479.3:p.(Gly217\*) | Homo | Hughes, 2019 | PMDS |
| 6 | Turkey | 5 | NM\_000479.3:c.1577G>T | NM\_000479.3:p.(Cys526Phe) | Homo | Bugrul, 2020 | PMDS |
| 7 | Turkey | 5 | NM\_000479.3:c.1673G>A | NM\_000479.3:p.(Gly558Asp) | Homo | Bugrul, 2020 | PMDS |
|  |  |  |  |  |  |  |  |
| 9 | Europe | 1 | NM\_000479.3:c.295A>T | NM\_000479.3:p.(Thr99Ser) | Hetero (1 allele) | Malone, 2019 | HH |
| 10 | Europe | 2 | NM\_000479.3:c.451C>T | NM\_000479.3:p.(Pro151Ser) | Hetero (1 allele) | Malone, 2019 | HH |
| 11 | Europe | 4 | NM\_000479.3:c.714C>A | NM\_000479.3:p.(Asp238Glu) | Hetero (1 allele) | Malone, 2019 | HH |

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**Supplementary Table 2.** Variants in the human *AMHR2* gene published after the exhaustive review by Picard et al., 2017.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient** | **Origin** | **Exon-intron** | **Variant** | **Effect** | **Alleles** | **Publication** | **Condition** |
| 1 | Turkey | 1 | NM\_020547.2:c.24G>A | NM\_020547.2:p.(Trp8\*) | Homo | Ata, 2021 | PMDS |
| 2 | India | 2 | NM\_020547.2:c.119G>C | NM\_020547.2:p.Gly40Ala | Homo | Fernandez Cancio, 2019 | PMDS |
| 3 | Turkey | 2nd Intr | NM\_020547.2:c.233–1G>A |  | Homo | Unal, 2018 | PMDS |
| 4 | ? | 3 | NM\_020547.2:c.237C>G | NM\_020547.2:p.Cys79Trp | Homo | Eggers, 2016 | PMDS |
| 5 | ? | 3 | NM\_020547.2:c.289C>T | NM\_020547.2:p.(Arg97\*) | Homo | Hughes, 2019 | PMDS |
| 6 | Spain | 4 | NM\_020547.2:c.502G>A | NM\_020547.2:p.Ala168Thr | Hetero (compound) | Orós-Millán, 2017 | PMDS |
| 6 | Spain | 7 | NM\_020547.2:c.877C>T | NM\_020547.2:p.Gln293\* | Hetero (compound) | Orós-Millán, 2017 | PMDS |
| 7 | ? | 6 | NM\_020547.2:c.745C>T | NM\_020547.2:p.Leu249Phe | Hetero (compound) | Baxter, 2015 | PMDS |
| 7 | ? | 10 | NM\_020547.2:c.1332\_1358del | NM\_020547.2:p.Gly445\_Leu453del | Hetero (compound) | Baxter, 2015 | PMDS |
| 8 | ? | 6 | NM\_020547.2:c.813\_817delGCTCT | NM\_020547.2:p.Leu272Trpfs\*24 | Hetero (compound) | Hughes, 2019 | PMDS |
| 8 | ? | 7 | NM\_020547.2:c.931G>A | NM\_020547.2:p.(Gly311Ser) | Hetero (compound) | [Hughes, 2019](https://www.ncbi.nlm.nih.gov/pubmed/?term=Elias-Assad) | PMDS |
| 9 | Colombia | 7 | NM\_020547.2:c.916delC | NM\_020547.2:p.Leu306Cysfs\*29 | Homo | García Acero, 2019 | PMDS |
| 10 | China | 9 | NM\_020547.2:c.1186\_1187delC | NM\_020547.2:p.Leu396Glyfs\*13 | Hetero (compound) | Ren, 2017 | PMDS |
| 10 | China | 10 | NM\_020547.2:c.1388G>A | NM\_020547.2:p.Arg463His | Hetero (compound) | Ren, 2017 | PMDS |
| 11 | ? | 10 | NM\_020547.2:c.1332\_1358del | NM\_020547.2:p.Gly445\_Leu453del | Hetero (compound) | Baxter, 2015 | PMDS |
| 11 | ? | 10 | NM\_020547.2:c.1343C>G | NM\_020547.2:p.Pro448Arg | Hetero (compound) | Baxter, 2015 | PMDS |
| 12 | Turkey | 10 | NM\_020547.2:c.1372G>T | NM\_020547.2:p.Val458Leu | Homo | Çakir, 2017 | PMDS |
| 13 | Turkey | 11 | NM\_020547.2:c.1510C>T | NM\_020547.2:p.Arg504Cys | Homo | Bugrul, 2020 | PMDS |
| 14 | Romania |  | del 12q13.13 chr12:53,829,597-53,833,931 | 11,39 kb, removing exons 7–11 and extending far beyond the AMHR2 gene | Homo | Tosca, 2020 | PMDS |
| 15 | Netherlands |  | del 12q13.13 | 10,53 kb, encompassing the whole AMHR2 gene | Hetero (compound) | Tosca, 2020 | PMDS |
|  |  |  |  |  |  |  |  |
| 21 | China | 6 | NM\_020547.2:c.626T>A | NM\_020547.2:p.Ile209Asn | Hetero (1 allele) | Qin, 2014 | POI |
| 22 | China | 8 | NM\_020547.2:c.1060C>T | NM\_020547.2:p.Lys354Phe | Hetero (1 allele) | Qin, 2014 | POI |
|  |  |  |  |  |  |  |  |
| 23 | European | 10 | NM\_020547.2:c.1330\_1356del | NM\_020547.2:p.Gly445\_Leu453del | Hetero (1 allele) | Malone, 2019 | HH |

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