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| Patient | Gene | Assembly ID | Transcript ID | Chromosomal location | rs | Mutation | a.a. change | Exon-intron | variant type | Type | Parental Validation | Mutation type | Evidence level | Variant classification |
| 1 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251795-2251797 | None | c.1522\_1524dupGTG | p.V508dup | Exon 5 | in-frame | Het | F | Novel | PM2 PP1 PP4 | VUS |
| chr19:2251910 | rs748647281 | c.1637C>A | p.A546E | Exon 5 | missense | Het | M | Known | PM2 PP1 PP4 | VUS |
| 2 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251795-2251797 | None | c.1522\_1524dupGTG | p.V508dup | Exon 5 | in-frame | Het | F | Novel | PM2 PP1 PP4 | VUS |
| chr19:2251910 | rs748647281 | c.1637C>A | p.A546E | Exon 5 | missense | Het | M | Known | PM2 PP1 PP4 | VUS |
| 3 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251625 | rs1235377959 | c.1352G>A | p.R451H | Exon 5 | missense | Het | F | Novel | PM2 PM3 PP4 | VUS |
| chr19:2249632 | rs778071215 | c.301G>A | p.G101R | Exon 1 | missense | Het | M | Known | PM2 PP1\_strong PP3 PP4 | LP |
| 4 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251877 | None | c.1604T>C | p.L535P | Exon 5 | missense | Het | F | Novel | PM2 PP3 PP4 | VUS |
| chr19:2250679 | None | c.584A>G | p.Y195C | Exon 3 | missense | Het | M | Novel | PM2 PP4 | VUS |
| 5 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251438 | rs1415701260 | c.1165G>T | p.E389X | Exon 5 | nonsense | Het | F | Known | PM2 PM3 PM4 PP1 PP3 PP4 | P |
| chr19:2114728-2456964 | None | \ | \ | Whole gene | Gross deletion | Het | M | Novel | PVS1 PM2 PP4 | P |
| 6 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2250890 | None | c.707G> A | p.G236D | Exon 4 | missense | Het | F | Novel | PM2 PM3 PP3 PP4 | LP |
| chr19:2249632 | rs778071215 | c.301G> A | p.G101R | Exon 1 | missense | Het | M | Known | PM2 PP1\_strong PP3 PP4 | LP |
| 7 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19: 2251438 | rs1415701260 | c.1165G>T | p.E389X | Exon 5 | nonsense | Hom | F | Known | PVS1 PM2 PP1 PP3 PP4 | P |
| chr19: 2251438 | rs1415701260 | c.1165G>T | p.E389X | Exon 5 | nonsense | Hom | M | Known | PVS1 PM2 PP1 PP3 PP4 | P |
| 8 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251265 | None | c.992C>T | p.S331L | Exon 5 | missense | Het | F | Novel | PM2 PM3 PP3 PP4 | LP |
| chr19:2249632 | rs778071215 | c.301G>A | p.G101R | Exon 1 | missense | Het | M | Known | PM2 PP1\_strong PP3 PP4 | LP |
| 9 | AMH | GRCh37/hg19 | NM\_000479.3 | chr19:2251720 | rs764585665 | c.1447T>C | p.Y483H | Exon 5 | missense | Het | F | Novel | PM2 PM3 PP3 PP4 | LP |
| chr19: 2249433 | None | c.102dupC | p.S35Qfs\*46 | Exon 1 | frameshift | Het | M | Novel | PVS1 PM2 PP4 | P |
| 10 | AMHR2 | GRCh37/hg19 | NM\_020547.3 | chr12:53818616 | None | c.356A>G | p.N119S | Exon 3 | missense | Het | F | Novel | PM2 PP3 PP4 | VUS |
| chr12:53819584 | rs1439647673 | c.733G>A | p.A245T | Exon 6 | missense | Het | M | Novel | PM2 PP4 | VUS |
| 11 | AMHR2 | GRCh37/hg19 | NM\_020547.3 | chr12:53819557 | None | c.706T>A | p.S236T | Exon 6 | missense | Het | F | Novel | PM2 PM3 PP4 | VUS |
| chr12:53818182 | rs534999427 | c.160C>T | p.R54C | Exon 2 | missense | Het | M | Known | PS3 PM2 PP3 PP4 | LP |
| 12 | AMHR2 | GRCh37/hg19 | NM\_020547.3 | chr12:53818597 | None | c.337A>C | p.T113P | Exon 3 | missense | Het | M | Novel | PM2 PP3 PP4 | VUS |
| chr12:53823785 | rs571389839 | c.1288+23C>A | ? | Intron 9 | missense | Het | F | Novel | PM2 PP4 | VUS |