|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| AMH | c.1762G>A | rs111822484 | - | 515 | Ala>Thr | NA |
| NM\_000479.3 | c.1771G>A | rs144513424 | - | 518 | Ala>Thr | NA |
| NP\_000470.2 | c.1793G>A | - | VAR\_031028 | 525 | Cys>Tyr | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.1795T>A | rs112266378 | - | 526 | Cys>Ser | NA |
|  | c.535G>A | rs182295886 | - | 106 | Gly>Ser | NA |
|  | c.569G>A | rs185020288 | - | 117 | Arg>Gln | NA |
|  | c.1649T>C | - | VAR\_007492 | 477 | Val>Ala | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.242G>C | rs145122767 | - | 8 | Ser>Thr | NA |
|  | c.428T>C | - | VAR\_007485 | 70 | Leu>Pro | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.1193A>G | rs140765565 VAR\_007491 | | 325 | Gln>Arg | NA |
|  | c.1737C>G | rs138571039 VAR\_031027 | | 506 | His>Gln | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.239C>G | rs149953700 | - | 7 | Thr>Ser | NA |
|  | c.254T>G | - | VAR\_007483 | 12 | Val>Gly | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.719A>G | - | VAR\_007488 | 167 | Tyr>Cys | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.772C>G | - | VAR\_007489 | 185 | Gln>Glu | NA |
|  | c.1607C>T | rs146124583 | - | 463 | Ala>Val | NA |
|  | c.1763C>T | rs10417628 | VAR\_065100 | 515 | Ala>Val | NA |
|  | c.272C>T | rs61736578 | - | 18 | Ala>Val | NA |
|  | c.365G>T | rs10407022 | VAR\_007484 | 49 | Ser>Ile | NA |
|  | c.521G>T | - | VAR\_007486 | 101 | Gly>Val | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.586C>T | - | VAR\_007487 | 123 | Arg>Trp | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  | c.647C>T | rs139265145 | - | 143 | Thr>Ile | NA |
|  | c.799C>T | - | VAR\_007490 | 194 | Arg>Cys | Persistent Muellerian duct syndrome type 1 (PMDS1) MIM:261550 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| Srd5a2 | c.216G>A | rs9282858 | VAR\_013104 | 49 | Ala>Thr | NA |
| NM\_000348.3 | c.222G>A | rs61748123 | - | 51 | Ala>Thr | NA |
| NP\_000339.2 | c.235T>A | rs121434245 VAR\_013105 | | 55 | Leu>Gln | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.280G>A | rs61750387 | - | 70 | Arg>Gln | NA |
|  | c.415G>A | rs121434246 | VAR\_013106 | 115 | Gly>Asp | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.438G>A | - | VAR\_025854 | 123 | Gly>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.543G>A | - | VAR\_025856 | 158 | Gly>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.618G>A | rs121434247 VAR\_013108 | | 183 | Gly>Ser | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.657G>A | rs121434250 | VAR\_013109 | 196 | Gly>Ser | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.669G>A | - | VAR\_013132 | 200 | Glu>Lys | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.678G>A | rs9332961 | VAR\_059791 | 203 | Gly>Ser | NA |
|  | c.691C>A | - | VAR\_025857 | 207 | Ala>Asp | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.742T>A | rs9332963 | VAR\_059792 | 224 | Leu>His | NA |
|  | c.751G>A | rs9332964 | VAR\_037586 | 227 | Arg>Gln | NA |
|  | c.753G>A | rs121434249 | VAR\_013112 | 228 | Ala>Thr | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.805C>A | - | VAR\_013133 | 245 | Ser>Tyr | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.808G>A | rs9332967 | VAR\_013134 | 246 | Arg>Gln | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.438G>C | - | VAR\_025854 | 123 | Gly>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.543G>C | - | VAR\_025856 | 158 | Gly>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.621G>C | rs61750398 | - | 184 | Gly>Arg | NA |
|  | c.84T>C | rs61748120 | - | 5 | Cys>Arg | NA |
|  | c.214C>G | rs61748122 | - | 48 | Pro>Arg | NA |
|  | c.336C>G | rs523349 | VAR\_013131 | 89 | Leu>Val | NA |
|  | c.408C>G | rs28383048 | VAR\_022302 | 113 | Leu>Val | NA |
|  | c.448A>G | - | VAR\_025855 | 126 | Gln>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.653C>G | rs61748126 | - | 194 | Phe>Leu | NA |
|  | c.706C>G | rs121434252 VAR\_013111 | | 212 | Pro>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.763A>G | rs121434251 VAR\_013113 | | 231 | His>Arg | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.773C>G | rs9332966 | - | 234 | Phe>Leu | NA |
|  | c.285C>T | rs61748124 | - | 72 | Pro>Ser | NA |
|  | c.504C>T | - | VAR\_025851 | 145 | Arg>Trp | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.613C>T | - | VAR\_025852 | 181 | Pro>Leu | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.631C>T | rs61748125 | - | 187 | Thr>Met | NA |
|  | c.662G>T | rs121434253 VAR\_013110 | | 197 | Glu>Asp | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.715C>T | rs34552434 | - | 215 | Ala>Val | NA |

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|  | c.775A>T | - | VAR\_025853 | 235 | Tyr>Phe | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  | c.807C>T | rs121434244 VAR\_005609 | | 246 | Arg>Trp | Pseudovaginal perineoscrotal hypospadias (PPSH) MIM:264600 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| SOX9 | c.1808C>A | - | - | 479 | Pro>His | NA |
| NM\_000346.3 | c.599C>A | rs137853128 VAR\_063642 | | 76 | Ala>Glu | Campomelic dysplasia (CMD1) MIM:114290 |
| NP\_000337.1 | c.708C>A | - | VAR\_003736 | 112 | Phe>Leu | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.799T>A | - | VAR\_003739 | 143 | Trp>Arg | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.844G>A | rs137853130 VAR\_008530 | | 158 | Ala>Thr | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.867C>A | - | VAR\_063645 | 165 | His>Gln | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.1156G>C | rs150378213 | - | 262 | Gly>Arg | NA |
|  | c.1163G>C | - | - | 264 | Arg>Thr | NA |
|  | c.1286G>C | rs143697828 | - | 305 | Gly>Ala | NA |
|  | c.559G>C | rs149852681 | - | 63 | Glu>Gln | NA |
|  | c.706T>C | - | VAR\_003736 | 112 | Phe>Leu | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.707T>C | - | VAR\_003737 | 112 | Phe>Ser | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.710T>C | - | VAR\_063643 | 113 | Met>Thr | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.799T>C | - | VAR\_003739 | 143 | Trp>Arg | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.827G>C | - | VAR\_003740 | 152 | Arg>Pro | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.940G>C | rs61740803 | - | 190 | Glu>Gln | NA |
|  | c.1351A>G | rs76093568 | - | 327 | Thr>Ala | NA |
|  | c.1490C>G | rs112211472 | - | 373 | Pro>Arg | NA |
|  | c.1882A>G | rs149888060 | - | 504 | Thr>Ala | NA |
|  | c.659A>G | rs148407362 | - | 96 | Asn>Ser | NA |
|  | c.708C>G | - | VAR\_003736 | 112 | Phe>Leu | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.709A>G | - | VAR\_063644 | 113 | Met>Val | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.834C>G | rs137853129 VAR\_008529 | | 154 | Phe>Leu | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.867C>G | - | VAR\_063645 | 165 | His>Gln | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.875A>G | - | - | 168 | Asp>Gly | NA |
|  | c.881C>G | - | VAR\_003741 | 170 | Pro>Arg | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.889A>G | rs104894647 VAR\_063647 | | 173 | Lys>Glu | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.1133G>T | rs183824168 | - | 254 | Arg>Leu | NA |
|  | c.1157G>T | rs138685561 | - | 262 | Gly>Val | NA |
|  | c.544C>T | - | - | 58 | Pro>Ser | NA |
|  | c.695C>T | - | VAR\_003735 | 108 | Pro>Leu | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.728C>T | - | VAR\_003738 | 119 | Ala>Val | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.865C>T | rs28940282 | VAR\_008531 | 165 | His>Tyr | Campomelic dysplasia (CMD1) MIM:114290 |
|  | c.881C>T | - | VAR\_063646 | 170 | Pro>Leu | Campomelic dysplasia (CMD1) MIM:114290 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| FGFR1 | c.1084G>A | rs121909640 VAR\_030968 | | 48 | Gly>Ser | Idiopathic hypogonadotropic hypogonadism (IHH) MIM:146110 |
| NM\_001174063.1 | c.1103G>A | rs145315779 | - | 54 | Arg>His | NA |
| NP\_001167534.1 | c.1150G>A | rs140254426 | - | 70 | Gly>Arg | NA |
|  | c.1173C>A | - | VAR\_030969 | 77 | Asn>Lys | NA |
|  | c.1232G>A | - | VAR\_017885 | 97 | Gly>Asp | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1246G>A | rs55642501 | VAR\_030972 | 102 | Val>Ile | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1339G>A | - | - | 133 | Asp>Asn | NA |
|  | c.1474T>A | - | VAR\_030974 | 178 | Cys>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1651G>A | rs121909635 VAR\_030978 | | 237 | Gly>Ser | NA |
|  | c.1652G>A | - | VAR\_030977 | 237 | Gly>Asp | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1691G>A | rs121909645 | - | 250 | Arg>Gln | NA |
|  | c.1696C>A | rs121913472 VAR\_042202 | | 252 | Pro>Thr |  |
|  | c.1703G>A | - | VAR\_030981 | 254 | Arg>Gln | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1751G>A | - | VAR\_030982 | 270 | Gly>Asp | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1759G>A | - | VAR\_030983 | 273 | Val>Met | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1772G>A | - | VAR\_017888 | 277 | Cys>Tyr | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.1886G>A | - | - | 315 | Gly>Glu | NA |
|  | c.2494G>A | - | VAR\_030995 | 518 | Ala>Thr | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.2574C>A | - | - | 544 | Asn>Lys | NA |
|  | c.2755G>A | rs121909629 VAR\_017889 | | 605 | Val>Met | Kallmann syndrome type 2 (KAL2) MIM:147950 |
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| c.2801G>A | - | VAR\_030999 | 620 | Arg>Gln | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2932T>A | - | VAR\_017890 | 664 | Trp>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2995G>A | - | VAR\_031001 | 685 | Gly>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3043G>A | - | VAR\_031004 | 701 | Gly>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3101C>A | - | VAR\_031005 | 720 | Pro>His | Idiopathic hypogonadotropic hypogonadism (IHH) MIM:146110 |
| c.3108C>A | - | VAR\_031007 | 722 | Asn>Lys | Idiopathic hypogonadotropic hypogonadism (IHH) MIM:146110 |
| c.3319G>A | - | VAR\_031010 | 793 | Val>Ile | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3388G>A | rs17182456 | VAR\_019291 | 816 | Gly>Arg | NA |
| c.1008G>C | rs17175750 | VAR\_019290 | 22 | Arg>Ser | NA |
| c.1325A>C | rs77734798 | - | 128 | Asp>Ala | NA |
| c.1328A>C | - | VAR\_030973 | 129 | Asp>Ala | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1475G>C | - | VAR\_030974 | 178 | Cys>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1612G>C | - | VAR\_030976 | 224 | Asp>His | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1676T>C | - | VAR\_030979 | 245 | Leu>Pro | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1725G>C | rs4647901 | - | 261 | Leu>Phe | NA |
| c.1841T>C | rs121909633 VAR\_030986 | | 300 | Ile>Thr | Trigonocephaly non-syndromic (TRICEPH) MIM:190440 |
| c.1865T>C | rs1126485 | - | 308 | Val>Ala | NA |
| c.1967T>C | rs121909638 | - | 342 | Leu>Ser | NA |
| c.2083T>C | rs121909634 | VAR\_030994 | 381 | Cys>Arg | Osteoglophonic dysplasia (OGD) MIM:166250 |
| c.2926G>C | - | VAR\_042203 | 662 | Val>Leu |  |
| c.2932T>C | - | VAR\_017890 | 664 | Trp>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2995G>C | - | VAR\_031001 | 685 | Gly>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3043G>C | - | VAR\_031003 | 701 | Gly>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1173C>G | - | VAR\_030969 | 77 | Asn>Lys | NA |
| c.1238A>G | - | VAR\_017886 | 99 | Tyr>Cys | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1364C>G | - | - | 141 | Thr>Arg | NA |
| c.1579T>G | rs17851623 | VAR\_030975 | 213 | Trp>Gly | NA |
| c.1697C>G | rs121909627 | VAR\_004111 | 252 | Pro>Arg | Pfeiffer syndrome (PS) MIM:101600 |
| c.1763A>G | - | VAR\_030984 | 274 | Glu>Gly | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1790C>G | - | VAR\_030985 | 283 | Pro>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1937C>G | - | VAR\_030988 | 332 | Ser>Cys | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1958A>G | - | VAR\_030989 | 339 | Tyr>Cys | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1979C>G | - | VAR\_030991 | 346 | Ser>Cys | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2063A>G | rs121909631 VAR\_030993 | | 374 | Tyr>Cys | Osteoglophonic dysplasia (OGD) MIM:166250 |
| c.2203A>G | - | - | 421 | Ile>Val | NA |
| c.2474T>G | rs77988343 | - | 511 | Val>Gly | NA |
| c.2548A>G | - | VAR\_030996 | 536 | Ile>Val | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2633A>G | - | - | 564 | Lys>Arg | NA |
| c.2798A>G | - | VAR\_030997 | 619 | His>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2800C>G | - | VAR\_030998 | 620 | Arg>Gly | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2902A>G | - | - | 654 | Lys>Glu | NA |
| c.3092T>G | - | VAR\_017891 | 717 | Met>Arg | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3108C>G | - | VAR\_031007 | 722 | Asn>Lys | Idiopathic hypogonadotropic hypogonadism (IHH) MIM:146110 |
| c.3241C>G | rs2956723 | VAR\_031009 | 767 | Leu>Val | NA |
| c.1010C>T | rs143341876 | - | 23 | Pro>Leu | NA |
| c.1153G>T | rs146670848 | - | 71 | Val>Leu | NA |
| c.1174C>T | - | VAR\_030970 | 78 | Arg>Cys | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1204G>T | rs139867599 | - | 88 | Val>Leu | NA |
| c.1244G>T | - | VAR\_030971 | 101 | Cys>Phe | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1316C>T | rs121913473 VAR\_042201 | | 125 | Ser>Leu |  |
| c.1390C>T | - | - | 150 | Pro>Ser | NA |
| c.1441G>T | rs121909630 VAR\_017887 | | 167 | Ala>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1690C>T | - | VAR\_030980 | 250 | Arg>Trp | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.1931A>T | rs121909632 | VAR\_030987 | 330 | Asn>Ile | Osteoglophonic dysplasia (OGD) MIM:166250 |
| c.1970C>T | - | VAR\_030990 | 343 | Ala>Val | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.2039C>T | rs121909641 VAR\_030992 | | 366 | Pro>Leu | NA |
| c.2234C>T | - | - | 431 | Ser>Phe | NA |
| c.2345G>T | rs121909637 | - | 468 | Arg>Leu | NA |
| c.2662C>T | - | - | 574 | Arg>Trp | NA |
| c.2926G>T | - | VAR\_042203 | 662 | Val>Leu |  |
| c.2990C>T | - | VAR\_031000 | 683 | Ser>Phe | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3013A>T | - | VAR\_031002 | 691 | Ile>Phe | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3100C>T | rs121909642 VAR\_031006 | | 720 | Pro>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |
| c.3169C>T | - | VAR\_031008 | 743 | Pro>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |

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|  | c.3228G>T | rs121909643 | - | 762 | Gln>His | NA |
|  | c.3238G>T | rs121909644 | - | 766 | Asp>Tyr | NA |
|  | c.3250C>T | rs56234888 | VAR\_017892 | 770 | Pro>Ser | Kallmann syndrome type 2 (KAL2) MIM:147950 |
|  | c.3400C>T | rs17182463 | VAR\_019292 | 820 | Arg>Cys | NA |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| AR | c.1119G>A | rs104894742 | - | 2 | Glu>Lys | NA |
| NM\_000044.2 | c.1234G>A | rs147842041 | - | 40 | Arg>Lys | NA |
| NP\_000035.2 | c.1285T>A | rs78686797 | VAR\_004681 | 57 | Leu>Gln |  |
|  | c.1386C>A | rs112374098 | - | 91 | Gln>Lys | NA |
|  | c.1736C>A | - | VAR\_009714 | 207 | Ser>Arg | NA |
|  | c.1761G>A | - | VAR\_009715 | 216 | Gly>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.1980G>A | - | - | 289 | Glu>Lys | NA |
|  | c.1984G>A | - | - | 290 | Cys>Tyr | NA |
|  | c.2140C>A | rs138454018 | - | 342 | Pro>Gln | NA |
|  | c.2178G>A | - | - | 355 | Glu>Lys | NA |
|  | c.2589G>A | - | VAR\_009719 | 492 | Gly>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2794G>A | - | VAR\_009723 | 560 | Cys>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2836C>A | - | VAR\_009728 | 574 | Ala>Asp | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2847G>A | rs137852596 | - | 578 | Gly>Arg | NA |
|  | c.2854G>A | - | VAR\_009734 | 580 | Cys>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2863T>A | rs137852587 | VAR\_009738 | 583 | Phe>Tyr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.2872G>A | - | VAR\_009740 | 586 | Arg>Lys | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2904G>A | rs137852569 VAR\_009743 | | 597 | Ala>Thr | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2938G>A | rs137852573 VAR\_004684 | | 608 | Arg>Gln | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.2941G>A | rs137852576 | VAR\_004685 | 609 | Arg>Lys | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.2950G>A | - | VAR\_009749 | 612 | Cys>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.2962G>A | - | VAR\_009751 | 616 | Arg>His | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.2974G>A | - | VAR\_009756 | 620 | Cys>Tyr |  |
|  | c.3004G>A | - | VAR\_009757 | 630 | Arg>Gln |  |
|  | c.3039G>A | rs111468555 | - | 642 | Glu>Lys | NA |
|  | c.3052C>A | rs1800053 | VAR\_004686 | 646 | Ala>Asp | NA |
|  | c.3058G>A | rs137852584 VAR\_009760 | | 648 | Ser>Asn |  |
|  | c.3109T>A | - | VAR\_004687 | 665 | Ile>Asn | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3130C>A | - | VAR\_009762 | 672 | Pro>His | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3159G>A | - | VAR\_009764 | 682 | Glu>Lys | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3162C>A | - | VAR\_013474 | 683 | Pro>Thr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3168G>A | - | VAR\_009766 | 685 | Val>Ile | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3181G>A | - | VAR\_009769 | 689 | Gly>Glu | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3201G>A | - | VAR\_004691 | 696 | Asp>Asn | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3216T>A | - | VAR\_009771 | 701 | Leu>Met | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3220T>A | - | VAR\_009773 | 702 | Leu>His | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3261G>A | - | VAR\_009781 | 716 | Val>Met |  |
|  | c.3279G>A | rs137852583 VAR\_009784 | | 722 | Ala>Thr |  |
|  | c.3285C>A | - | - | 724 | Pro>Thr | NA |
|  | c.3289G>A | - | VAR\_009787 | 725 | Gly>Asp | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3293C>A | - | VAR\_009788 | 726 | Phe>Leu | NA |
|  | c.3299C>A | - | VAR\_009790 | 728 | Asn>Lys | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3300T>A | - | - | 729 | Leu>Ile | NA |
|  | c.3306G>A | rs137852571 VAR\_004695 | | 731 | Val>Met |  |
|  | c.3312G>A | - | VAR\_004696 | 733 | Asp>Asn | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3339T>A | - | VAR\_009794 | 742 | Trp>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3344G>A | - | VAR\_004698 | 743 | Met>Ile | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3346G>A | rs137852600 | VAR\_013477 | 744 | Gly>Glu | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3354G>A | - | VAR\_009798 | 747 | Val>Met | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3360G>A | - | VAR\_009800 | 749 | Ala>Thr |  |
|  | c.3361C>A | - | VAR\_009799 | 749 | Ala>Asp | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3365G>A | - | VAR\_009802 | 750 | Met>Ile |  |
|  | c.3366G>A | - | VAR\_009803 | 751 | Gly>Ser |  |
|  | c.3367G>A | - | VAR\_004701 | 751 | Gly>Asp | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3369T>A | - | VAR\_009804 | 752 | Trp>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3373G>A | - | VAR\_004702 | 753 | Arg>Gln | Androgen insensitivity syndrome (AIS) MIM:300068 |

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| c.3380C>A | - | VAR\_009805 | 755 | Phe>Leu | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3410C>A | - | VAR\_009813 | 765 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3411G>A | - | VAR\_004707 | 766 | Ala>Thr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3419T>A | - | VAR\_009816 | 768 | Asp>Glu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3439G>A | rs137852572 | VAR\_004708 | 775 | Arg>His | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3463G>A | - | VAR\_009821 | 783 | Ser>Asn |  |
| c.3469G>A | - | VAR\_004712 | 785 | Cys>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3535G>A | - | VAR\_009826 | 807 | Cys>Tyr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3559G>A | - | VAR\_004718 | 815 | Ser>Asn | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3610G>A | - | VAR\_004720 | 832 | Arg>Gln | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3633G>A | - | - | 840 | Asp>Asn | NA |
| c.3636C>A | - | VAR\_009229 | 841 | Arg>Ser | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3637G>A | rs9332969 | VAR\_004723 | 841 | Arg>His | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3679G>A | - | VAR\_009835 | 855 | Arg>Lys | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3682G>A | rs9332971 | VAR\_004726 | 856 | Arg>His | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3708G>A | - | VAR\_004727 | 865 | Asp>Asn | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3714G>A | rs137852564 | VAR\_004730 | 867 | Val>Met | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3715T>A | - | VAR\_004728 | 867 | Val>Glu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3757T>A | - | VAR\_009845 | 881 | Leu>Gln |  |
| c.3783G>A | - | VAR\_009848 | 890 | Val>Met | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3786G>A | - | VAR\_009849 | 891 | Asp>Asn |  |
| c.3791T>A | - | VAR\_009850 | 892 | Phe>Leu |  |
| c.3804G>A | - | VAR\_009851 | 897 | Ala>Thr |  |
| c.3825G>A | - | VAR\_009854 | 904 | Val>Met | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3829C>A | - | VAR\_009855 | 905 | Pro>His | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3843G>A | - | VAR\_009858 | 910 | Gly>Arg | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3844G>A | - | VAR\_009857 | 910 | Gly>Glu |  |
| c.3866C>A | - | VAR\_009861 | 917 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.1276T>C | - | VAR\_004680 | 54 | Leu>Ser |  |
| c.1457G>C | - | VAR\_009712 | 114 | Gln>His |  |
| c.1734A>C | - | VAR\_009714 | 207 | Ser>Arg | NA |
| c.1761G>C | - | VAR\_009715 | 216 | Gly>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.1885T>C | - | VAR\_009225 | 257 | Leu>Pro | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.1918T>C | - | VAR\_009716 | 268 | Met>Thr |  |
| c.2759G>C | - | VAR\_009721 | 548 | Leu>Phe | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2839T>C | - | VAR\_009729 | 575 | Leu>Pro |  |
| c.2844T>C | - | VAR\_009732 | 577 | Cys>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2863T>C | - | VAR\_009737 | 583 | Phe>Ser | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2908G>C | - | VAR\_009745 | 598 | Ser>Thr | NA |
| c.2947A>C | - | VAR\_009748 | 611 | Asn>Thr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2962G>C | - | VAR\_009752 | 616 | Arg>Pro | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2965T>C | - | VAR\_009753 | 617 | Leu>Pro | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2968G>C | - | VAR\_009755 | 618 | Arg>Pro | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3007A>C | - | VAR\_009758 | 631 | Lys>Thr |  |
| c.3109T>C | rs9332968 | - | 665 | Ile>Thr | NA |
| c.3133T>C | - | VAR\_009763 | 673 | Ile>Thr |  |
| c.3148T>C | rs137852579 | VAR\_004688 | 678 | Leu>Pro | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3166G>C | - | VAR\_009765 | 684 | Gly>Ala |  |
| c.3174T>C | - | VAR\_009767 | 687 | Cys>Arg | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3184A>C | rs137852599 | - | 690 | His>Pro | NA |
| c.3201G>C | - | VAR\_004690 | 696 | Asp>His | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3241G>C | - | VAR\_009777 | 709 | Gly>Ala | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3247G>C | - | VAR\_009779 | 711 | Arg>Thr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3284G>C | - | VAR\_009785 | 723 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3291T>C | - | VAR\_009788 | 726 | Phe>Leu | NA |
| c.3301T>C | - | VAR\_009791 | 729 | Leu>Ser | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3317G>C | - | VAR\_009792 | 734 | Gln>His | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3328T>C | - | VAR\_009793 | 738 | Ile>Thr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3339T>C | - | VAR\_009794 | 742 | Trp>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3344G>C | - | VAR\_004698 | 743 | Met>Ile | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3352T>C | - | VAR\_009797 | 746 | Met>Thr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3365G>C | - | VAR\_009802 | 750 | Met>Ile |  |
| c.3369T>C | - | VAR\_009804 | 752 | Trp>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3378T>C | - | VAR\_009805 | 755 | Phe>Leu | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |

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| c.3388T>C | - | VAR\_009808 | 758 | Val>Ala |  |
| c.3391A>C | - | VAR\_009809 | 759 | Asn>Thr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3393T>C | - | VAR\_009811 | 760 | Ser>Pro |  |
| c.3405T>C | - | VAR\_009812 | 764 | Tyr>His | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3408T>C | - | VAR\_009813 | 765 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3421T>C | - | VAR\_009817 | 769 | Leu>Pro | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3429A>C | - | VAR\_009818 | 772 | Asn>His | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3433A>C | - | VAR\_009819 | 773 | Glu>Ala | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3482G>C | - | VAR\_009822 | 789 | Arg>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3489T>C | - | VAR\_009824 | 792 | Ser>Pro |  |
| c.3497G>C | - | VAR\_009825 | 794 | Glu>Asp | NA |
| c.3499T>C | - | VAR\_004714 | 795 | Phe>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3538T>C | rs137852592 | VAR\_009827 | 808 | Met>Thr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3577G>C | - | VAR\_009829 | 821 | Gly>Ala | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3607T>C | - | VAR\_009831 | 831 | Leu>Pro |  |
| c.3643T>C | rs9332970 | VAR\_004724 | 843 | Ile>Thr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3711T>C | rs137852597 | VAR\_009839 | 866 | Ser>Pro | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3789T>C | - | VAR\_009850 | 892 | Phe>Leu |  |
| c.3802T>C | - | VAR\_004734 | 896 | Met>Thr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3811T>C | - | VAR\_009852 | 899 | Ile>Thr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3843G>C | - | VAR\_009858 | 910 | Gly>Arg | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3849G>C | - | VAR\_009860 | 912 | Val>Leu | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3864T>C | - | VAR\_009861 | 917 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.1249C>G | rs139767835 | - | 45 | Ala>Gly | NA |
| c.1306A>G | - | VAR\_009711 | 64 | Gln>Arg |  |
| c.1660A>G | - | VAR\_009713 | 182 | Lys>Arg |  |
| c.1702A>G | - | VAR\_009224 | 196 | Gln>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.1736C>G | - | VAR\_009714 | 207 | Ser>Arg | NA |
| c.1820C>G | - | - | 235 | Asn>Lys | NA |
| c.2290C>G | - | VAR\_009226 | 392 | Pro>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2449A>G | - | VAR\_009228 | 445 | Gln>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2701A>G | - | VAR\_009720 | 529 | Asp>Gly |  |
| c.2830A>G | - | VAR\_009727 | 572 | Tyr>Cys | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2841A>G | - | VAR\_009730 | 576 | Thr>Ala |  |
| c.2857A>G | - | VAR\_009735 | 581 | Lys>Arg |  |
| c.2907A>G | - | VAR\_009744 | 598 | Ser>Gly | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2965T>G | - | VAR\_009754 | 617 | Leu>Arg | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3127A>G | - | VAR\_009761 | 671 | Gln>Arg |  |
| c.3132A>G | rs146618994 | - | 673 | Ile>Val | NA |
| c.3222T>G | - | VAR\_009774 | 703 | Ser>Ala | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3225A>G | - | VAR\_004693 | 704 | Ser>Gly | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3232A>G | - | VAR\_009776 | 706 | Asn>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3238T>G | rs137852585 | VAR\_004694 | 708 | Leu>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3249C>G | - | VAR\_013476 | 712 | Gln>Glu | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3267A>G | - | VAR\_009782 | 718 | Lys>Glu |  |
| c.3276A>G | - | VAR\_009783 | 721 | Lys>Glu |  |
| c.3293C>G | - | VAR\_009788 | 726 | Phe>Leu | NA |
| c.3299C>G | - | VAR\_009790 | 728 | Asn>Lys | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3337C>G | rs137852601 | - | 741 | Ser>Cys | NA |
| c.3342A>G | - | VAR\_009795 | 743 | Met>Val | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3363A>G | - | VAR\_004700 | 750 | Met>Val | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3378T>G | - | VAR\_004703 | 755 | Phe>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3380C>G | - | VAR\_009805 | 755 | Phe>Leu | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3381A>G | - | VAR\_009806 | 756 | Thr>Ala |  |
| c.3385A>G | rs141425171 VAR\_009807 | | 757 | Asn>Ser | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3406A>G | rs137852567 | VAR\_004705 | 764 | Tyr>Cys | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3410C>G | - | VAR\_009813 | 765 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3419T>G | - | VAR\_009816 | 768 | Asp>Glu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3433A>G | - | VAR\_009820 | 773 | Glu>Gly | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3477A>G | rs137852570 VAR\_004713 | | 788 | Met>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3510C>G | rs137852591 | VAR\_004715 | 799 | Gln>Glu | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3537A>G | - | VAR\_004717 | 808 | Met>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3538T>G | - | VAR\_004716 | 808 | Met>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3555T>G | - | - | 814 | Phe>Val | NA |

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| c.3579C>G | - | VAR\_009830 | 822 | Leu>Val | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3597T>G | - | VAR\_013478 | 828 | Phe>Val | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3619A>G | - | VAR\_009832 | 835 | Tyr>Cys | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3636C>G | - | VAR\_004722 | 841 | Arg>Gly | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3640T>G | - | VAR\_009833 | 842 | Ile>Ser | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3654A>G | - | VAR\_009834 | 847 | Arg>Gly |  |
| c.3686C>G | rs137852598 | VAR\_009836 | 857 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3706T>G | - | VAR\_009837 | 864 | Leu>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3709A>G | - | VAR\_009838 | 865 | Asp>Gly | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3725T>G | rs137852574 | VAR\_004731 | 870 | Ile>Met | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3727C>G | - | VAR\_009840 | 871 | Ala>Gly | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3729A>G | - | VAR\_009842 | 872 | Arg>Gly | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3739A>G | - | VAR\_013479 | 875 | His>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3747A>G | rs137852578 VAR\_004732 | | 878 | Thr>Ala |  |
| c.3748C>G | rs137852580 VAR\_009844 | | 878 | Thr>Ser |  |
| c.3759C>G | - | VAR\_009846 | 882 | Leu>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3774A>G | - | VAR\_009847 | 887 | Met>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3791T>G | - | VAR\_009850 | 892 | Phe>Leu |  |
| c.3823A>G | rs137852582 VAR\_009853 | | 903 | Gln>Arg |  |
| c.3847A>G | - | VAR\_009859 | 911 | Lys>Arg |  |
| c.3866C>G | - | VAR\_009861 | 917 | Phe>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3868A>G | - | VAR\_009862 | 918 | His>Arg | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3874A>G | - | VAR\_009863 | 920 | Gln>Arg |  |
| c.1297A>T | rs62636528 | - | 61 | Gln>Leu | NA |
| c.1303A>T | rs62636527 | - | 63 | Gln>Leu | NA |
| c.1376G>T | rs62636529 | - | 87 | Gln>His | NA |
| c.1457G>T | - | VAR\_009712 | 114 | Gln>His |  |
| c.1926C>T | - | VAR\_009717 | 271 | Pro>Ser |  |
| c.1929C>T | rs148972137 | - | 272 | Leu>Phe | NA |
| c.2140C>T | - | VAR\_009718 | 342 | Pro>Leu |  |
| c.2289C>T | - | VAR\_009227 | 392 | Pro>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2759G>T | - | VAR\_009721 | 548 | Leu>Phe | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2760C>T | rs137852588 | VAR\_009722 | 549 | Pro>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2820G>T | - | VAR\_009726 | 569 | Gly>Trp | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2821G>T | - | VAR\_009725 | 569 | Gly>Val | NA |
| c.2845G>T | - | VAR\_009731 | 577 | Cys>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2854G>T | rs137852586 | VAR\_009733 | 580 | Cys>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2859G>T | - | VAR\_009736 | 582 | Val>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2875C>T | - | VAR\_009741 | 587 | Ala>Val |  |
| c.2877G>T | - | VAR\_009742 | 588 | Ala>Ser |  |
| c.2920G>T | - | VAR\_009746 | 602 | Cys>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.2928G>T | - | VAR\_009747 | 605 | Asp>Tyr | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.2953C>T | - | - | 613 | Pro>Leu | NA |
| c.3178C>T | - | VAR\_009768 | 688 | Ala>Val | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3202A>T | - | VAR\_004692 | 696 | Asp>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3219C>T | - | VAR\_009772 | 702 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3225A>T | - | VAR\_009775 | 704 | Ser>Cys | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3231A>T | - | VAR\_013475 | 706 | Asn>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3241G>T | - | VAR\_009778 | 709 | Gly>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3252C>T | rs137852595 | VAR\_009780 | 713 | Leu>Phe | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3284G>T | - | VAR\_009785 | 723 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3285C>T | - | VAR\_009786 | 724 | Pro>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3295G>T | rs137852593 VAR\_009789 | | 727 | Arg>Leu |  |
| c.3312G>T | - | VAR\_004697 | 733 | Asp>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3317G>T | - | VAR\_009792 | 734 | Gln>His | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3344G>T | - | VAR\_004698 | 743 | Met>Ile | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3346G>T | rs137852600 | VAR\_004699 | 744 | Gly>Val | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
| c.3348C>T | - | VAR\_009796 | 745 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3361C>T | - | VAR\_009801 | 749 | Ala>Val |  |
| c.3365G>T | - | VAR\_009802 | 750 | Met>Ile |  |
| c.3394C>T | - | VAR\_009810 | 760 | Ser>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3402C>T | - | VAR\_004704 | 763 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3412C>T | - | VAR\_009814 | 766 | Ala>Val | Androgen insensitivity syndrome (AIS) MIM:300068 |
| c.3414C>T | - | VAR\_009815 | 767 | Pro>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |

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| --- | --- | --- | --- | --- | --- | --- |
|  | c.3438C>T | rs137852562 | VAR\_004709 | 775 | Arg>Cys | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3453C>T | - | VAR\_004710 | 780 | Arg>Trp | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3458G>T | rs137852589 | VAR\_004711 | 781 | Met>Ile | Androgen insensitivity syndrome (AIS) MIM:300068|Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3482G>T | - | VAR\_009822 | 789 | Arg>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3486C>T | - | VAR\_009823 | 791 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3497G>T | - | VAR\_009825 | 794 | Glu>Asp | NA |
|  | c.3552C>T | - | VAR\_009828 | 813 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3610G>T | - | VAR\_004719 | 832 | Arg>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3636C>T | rs137852577 | VAR\_004721 | 841 | Arg>Cys | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3681C>T | - | VAR\_004725 | 856 | Arg>Cys | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3714G>T | rs137852564 | VAR\_004729 | 867 | Val>Leu | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3727C>T | - | VAR\_009841 | 871 | Ala>Val | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  | c.3738C>T | rs137852581 | VAR\_009843 | 875 | His>Tyr |  |
|  | c.3753G>T | - | VAR\_013480 | 880 | Asp>Tyr | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3793C>T | - | VAR\_004733 | 893 | Pro>Leu | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3828C>T | - | VAR\_009856 | 905 | Pro>Ser | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3837C>T | - | VAR\_004735 | 908 | Leu>Phe | Androgen insensitivity syndrome (AIS) MIM:300068 |
|  | c.3855C>T | - | VAR\_004736 | 914 | Pro>Ser | Androgen insensitivity syndrome partial (PAIS) MIM:312300 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| SRY | c.201G>A | rs104894971 VAR\_003717 | | 18 | Ser>Asn | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
| NM\_003140.1 | c.340G>A | rs104894969 VAR\_003721 | | 64 | Met>Ile | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
| NP\_003131.1 | c.374C>A | - | VAR\_017300 | 76 | Arg>Ser | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.432G>A | rs104894972 | VAR\_017302 | 95 | Gly>Glu | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.450T>A | - | VAR\_003727 | 101 | Leu>His | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.485G>A | rs104894966 VAR\_003731 | | 113 | Ala>Thr | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.326G>C | rs104894957 | VAR\_003719 | 60 | Val>Leu | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.327T>C | - | VAR\_003718 | 60 | Val>Ala | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.351T>C | rs104894968 | VAR\_003722 | 68 | Ile>Thr | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.381T>C | - | VAR\_003723 | 78 | Met>Thr | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.431G>C | rs104894974 VAR\_003726 | | 95 | Gly>Arg | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.474T>C | rs104894956 | VAR\_003730 | 109 | Phe>Ser | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.332C>G | - | VAR\_003720 | 62 | Arg>Gly | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.339T>G | - | VAR\_017298 | 64 | Met>Arg | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.347T>G | - | VAR\_017299 | 67 | Phe>Val | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.418C>G | rs104894959 | VAR\_003724 | 90 | Ile>Met | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.419A>G | - | VAR\_003725 | 91 | Ser>Gly | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.471C>G | - | VAR\_003729 | 108 | Pro>Arg | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.528A>G | - | VAR\_003733 | 127 | Tyr>Cys | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.540C>G | - | VAR\_017304 | 131 | Pro>Arg | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.407A>T | - | VAR\_017301 | 87 | Asn>Tyr | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.465A>T | rs104894964 | VAR\_003728 | 106 | Lys>Ile | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.522C>T | - | VAR\_003732 | 125 | Pro>Leu | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.528A>T | rs104894973 VAR\_017303 | | 127 | Tyr>Phe | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.545C>T | rs104894976 | VAR\_003734 | 133 | Arg>Trp | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.471C>G | - | VAR\_003729 | 108 | Pro>Arg | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.528A>G | - | VAR\_003733 | 127 | Tyr>Cys | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.540C>G | - | VAR\_017304 | 131 | Pro>Arg | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.407A>T | - | VAR\_017301 | 87 | Asn>Tyr | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.465A>T | rs104894964 | VAR\_003728 | 106 | Lys>Ile | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.522C>T | - | VAR\_003732 | 125 | Pro>Leu | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.528A>T | rs104894973 VAR\_017303 | | 127 | Tyr>Phe | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  | c.545C>T | rs104894976 VAR\_003734 | | 133 | Arg>Trp | 46 XY sex reversal type 1 (SRXY1) MIM:400044 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| SRY | c.1085G>A | rs144981691 | - | 274 | Arg>His | NA |
| NM\_000349.2 | c.347T>A | - | - | 28 | Val>Glu | NA |
| NP\_000340.2 | c.374G>A | rs141136662 | - | 37 | Arg>Gln | NA |
|  | c.697G>A | rs80231229 | - | 145 | Gly>Arg | NA |
|  | c.769G>A | - | VAR\_014237 | 169 | Glu>Lys | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  |  |  |  |  |  |  |

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| --- | --- | --- | --- | --- | --- | --- |
|  | c.809G>A | rs104894086 | - | 182 | Arg>His | NA |
|  | c.823G>A | rs104894089 | - | 187 | Val>Met | NA |
|  | c.827G>A | rs61736315 | - | 188 | Arg>His | NA |
|  | c.872C>A | rs1042854 | VAR\_005628 | 203 | Ala>Asp | NA |
|  | c.925G>A | rs139081695 | - | 221 | Gly>Ser | NA |
|  | c.959G>A | rs148239025 | - | 232 | Gly>Glu | NA |
|  | c.1088T>C | - | VAR\_014242 | 275 | Leu>Pro | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  | c.280T>C | rs76755486 | - | 6 | Phe>Leu | NA |
|  | c.456A>C | rs77907027 | - | 64 | Glu>Asp | NA |
|  | c.914G>C | rs28938471 | VAR\_014238 | 217 | Arg>Thr | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  | c.938T>C | - | VAR\_014240 | 225 | Met>Thr | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  | c.955G>C | - | - | 231 | Ala>Pro | NA |
|  | c.770A>G | - | VAR\_014236 | 169 | Glu>Gly | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  | c.1052C>T | rs142623227 | - | 263 | Thr>Ile | NA |
|  | c.1084C>T | rs138161253 | - | 274 | Arg>Cys | NA |
|  | c.404C>T | rs182563252 | - | 47 | Thr>Met | NA |
|  | c.427C>T | rs151075160 | - | 55 | Arg>Trp | NA |
|  | c.625C>T | rs34908868 | VAR\_034520 | 121 | Arg>Trp | NA |
|  | c.809G>T | rs104894086 VAR\_005627 | | 182 | Arg>Leu | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  | c.826C>T | rs104894090 | - | 188 | Arg>Cys | NA |
|  | c.917C>T | rs137852690 VAR\_014239 | | 218 | Ala>Val | Adrenal hyperplasia type 1 (AH1) MIM:201710 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| NR0B1 | c.1048C>A | rs140259346 | - | 345 | Pro>Thr | NA |
| NM\_000475.4 | c.1144G>A | - | VAR\_004742 | 377 | Glu>Lys | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
| NP\_000466.2 | c.1157T>A | rs104894899 VAR\_018301 | | 381 | Leu>His | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1307G>A | - | - | 431 | Ser>Asn | NA |
|  | c.439C>A | - | - | 142 | Gln>Lys | NA |
|  | c.856G>A | - | - | 281 | Asp>Asn | NA |
|  | c.1289G>C | - | VAR\_018305 | 425 | Arg>Thr | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.330G>C | rs132630327 | - | 105 | Trp>Cys | NA |
|  | c.815G>C | rs104894888 | VAR\_004738 | 267 | Arg>Pro | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.848T>C | - | VAR\_031079 | 278 | Leu>Pro | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.888G>C | rs28935482 | VAR\_031080 | 291 | Trp>Cys | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.899T>C | - | VAR\_018303 | 295 | Leu>Pro | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.905T>C | rs104894907 | VAR\_031081 | 297 | Leu>Pro | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.913G>C | - | VAR\_018304 | 300 | Ala>Pro | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1153T>G | rs104894900 | VAR\_018300 | 380 | Tyr>Asp | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1169T>G | - | VAR\_004744 | 385 | Val>Gly | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1288A>G | - | VAR\_004745 | 425 | Arg>Gly | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1331T>G | rs104894897 | VAR\_018302 | 439 | Ile>Ser | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1412T>G | - | VAR\_018306 | 466 | Leu>Arg | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1417A>G | - | - | 468 | Thr>Ala | NA |
|  | c.1425A>G | rs151317312 | - | 470 | Ile>Met | NA |
|  | c.155C>G | rs61756004 | - | 47 | Pro>Arg | NA |
|  | c.875T>G | - | VAR\_004740 | 287 | Val>Gly | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1161G>T | rs104894896 VAR\_004743 | | 382 | Lys>Asn | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.1217A>T | rs11550590 | - | 401 | Gln>Leu | NA |
|  | c.1334A>T | rs28935481 | VAR\_004746 | 440 | Asn>Ile | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  | c.305C>T | rs139871029 | - | 97 | Pro>Leu | NA |
|  | c.850C>T | rs183613764 | - | 279 | Pro>Ser | NA |
|  | c.914C>T | - | VAR\_004741 | 300 | Ala>Val | X-linked adrenal hypoplasia congenital (XL-AHC) MIM:300200 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| WT1 | c.1062G>A | - | VAR\_007740 | 291 | Ser>Asn | Wilms tumor 1 (WT1) MIM:194070 |
| NM\_000378.4 | c.1161G>A | - | - | 324 | Ser>Asn | NA |
| NP\_000369.3 | c.1248G>A | - | - | 353 | Arg>His | NA |
|  | c.1278G>A | - | VAR\_015053 | 363 | Arg>Gln | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
|  | c.1332G>A | rs121907904 VAR\_007743 | | 381 | Cys>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
|  | c.1341G>A | - | - | 384 | Arg>Lys | NA |
|  |  |  |  |  |  |  |

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| --- | --- | --- | --- | --- | --- |
| c.1356C>A | - | - | 389 | Ser>Tyr | NA |
| c.1407G>A | - | VAR\_015055 | 406 | Cys>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1422G>A | - | VAR\_043800 | 411 | Cys>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1435T>A | - | VAR\_043801 | 415 | Phe>Leu | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1440G>A | rs121907901 VAR\_007746 | | 417 | Arg>His | Denys-Drash syndrome (DDS) MIM:194080|Wilms tumor 1 (WT1) MIM:194070 |
| c.1497G>A | - | - | 436 | Cys>Tyr | NA |
| c.1506G>A | - | VAR\_043806 | 439 | Cys>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1524G>A | - | VAR\_015061 | 445 | Arg>Gln | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1529G>A | rs28941778 | VAR\_007751 | 447 | Asp>Asn | Denys-Drash syndrome (DDS) MIM:194080|Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1534C>A | - | - | 448 | His>Gln | NA |
| c.1584G>A | rs139893274 | - | 465 | Arg>Gln | NA |
| c.713T>A | - | - | 175 | Cys>Ser | NA |
| c.785G>A | rs9332973 | VAR\_043798 | 199 | Ala>Thr | NA |
| c.996G>A | - | - | 269 | Gly>Asp | NA |
| c.1062G>C | - | - | 291 | Ser>Thr | NA |
| c.1185T>C | - | VAR\_058021 | 332 | Leu>Pro | NA |
| c.1187T>C | - | - | 333 | Cys>Arg | NA |
| c.1286T>C | - | - | 366 | Ser>Pro | NA |
| c.1322T>C | - | - | 378 | Tyr>His | NA |
| c.1355T>C | - | - | 389 | Ser>Pro | NA |
| c.1433T>C | - | VAR\_043801 | 415 | Phe>Leu | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1449A>C | - | VAR\_043803 | 420 | Gln>Pro | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1490T>C | rs28941777 | VAR\_007749 | 434 | Phe>Leu | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1496T>C | - | VAR\_015058 | 436 | Cys>Arg | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1505T>C | - | VAR\_043805 | 439 | Cys>Arg | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1517T>C | rs28941779 | VAR\_015060 | 443 | Phe>Leu | Frasier syndrome (FS) MIM:136680 |
| c.1524G>C | rs121907903 VAR\_043808 | | 445 | Arg>Pro | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1529G>C | - | - | 447 | Asp>His | NA |
| c.1533A>C | - | VAR\_043810 | 448 | His>Pro | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1536T>C | - | VAR\_015062 | 449 | Leu>Pro | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1551G>C | - | - | 454 | Arg>Thr | NA |
| c.855T>C | - | - | 222 | Phe>Ser | NA |
| c.1160A>G | rs121907908 VAR\_007742 | | 324 | Ser>Gly | NA |
| c.1200A>G | - | - | 337 | Tyr>Cys | NA |
| c.1207A>G | rs17855567 | - | 339 | Ile>Met | NA |
| c.1224T>G | rs150194429 | - | 345 | Phe>Cys | NA |
| c.1277C>G | - | - | 363 | Arg>Gly | NA |
| c.1281C>G | rs142937387 | - | 364 | Ser>Trp | NA |
| c.1349A>G | - | - | 387 | Lys>Glu | NA |
| c.1368T>G | - | VAR\_015054 | 393 | Met>Arg | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1380A>G | - | - | 397 | Lys>Arg | NA |
| c.1406T>G | - | VAR\_043799 | 406 | Cys>Gly | Wilms tumor 1 (WT1) MIM:194070 |
| c.1421T>G | rs121907905 VAR\_007744 | | 411 | Cys>Gly | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1430A>G | rs77183751 | - | 414 | Arg>Gly | NA |
| c.1435T>G | - | VAR\_043801 | 415 | Phe>Leu | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1455A>G | - | - | 422 | Lys>Arg | NA |
| c.1462C>G | rs121907907 VAR\_007747 | | 424 | His>Gln | Denys-Drash syndrome (DDS) MIM:194080|Wilms tumor 1 (WT1) MIM:194070 |
| c.1473A>G | - | VAR\_015057 | 428 | His>Arg | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1523C>G | - | - | 445 | Arg>Gly | NA |
| c.1530A>G | rs121907902 VAR\_007752 | | 447 | Asp>Gly | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1532C>G | - | - | 448 | His>Asp | NA |
| c.938A>G | rs142653301 | - | 250 | Met>Val | NA |
| c.1005C>T | rs138073760 | - | 272 | Thr>Ile | NA |
| c.1173C>T | - | - | 328 | Thr>Ile | NA |
| c.1212C>T | - | - | 341 | Thr>Met | NA |
| c.1238G>T | - | - | 350 | Asp>Tyr | NA |
| c.1439C>T | rs121907910 VAR\_007745 | | 417 | Arg>Cys | Denys-Drash syndrome (DDS) MIM:194080|Meacham syndrome (MEACHS) MIM:608978|Wilms tumor 1 (WT1) MIM:194070 |
| c.1440G>T | - | VAR\_043802 | 417 | Arg>Leu | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1460C>T | - | VAR\_015056 | 424 | His>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1472C>T | rs28942089 | VAR\_007748 | 428 | His>Tyr | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1478G>T | - | VAR\_043804 | 430 | Gly>Cys | Nephrotic syndrome type 4 (NPHS4) MIM:256370 |
| c.1479G>T | - | - | 430 | Gly>Val | NA |
| c.1506G>T | - | VAR\_015059 | 439 | Cys>Phe | Denys-Drash syndrome (DDS) MIM:194080 |
| c.1523C>T | rs121907900 VAR\_007750 | | 445 | Arg>Trp | Denys-Drash syndrome (DDS) MIM:194080|Meacham syndrome (MEACHS) MIM:608978|Wilms tumor 1 (WT1) MIM:194070 |

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|  | c.1524G>T | - | VAR\_043807 | 445 | Arg>Leu | Wilms tumor 1 (WT1) MIM:194070 |
|  | c.1529G>T | - | VAR\_043809 | 447 | Asp>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
|  | c.1544C>T | - | VAR\_043811 | 452 | His>Tyr | Denys-Drash syndrome (DDS) MIM:194080 |
|  | c.1557A>T | - | - | 456 | His>Leu | NA |
|  | c.1631C>T | - | - | 481 | Arg>Cys | NA |
|  | c.391G>T | rs5030135 | - | 67 | Gln>His | NA |
|  | c.486C>T | - | - | 99 | Ala>Val | NA |
|  | c.846C>T | - | - | 219 | Thr>Met | NA |
|  | c.935C>T | rs2234584 | VAR\_007739 | 249 | Pro>Ser | Wilms tumor 1 (WT1) MIM:194070 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| NR5A1 | c.1064G>A | rs121918655 VAR\_062970 | | 293 | Asp>Asn | Premature ovarian failure type 7 (POF7) MIM:612964 |
| NM\_004959.4 | c.1394G>A | rs150382425 | - | 403 | Asp>Asn | NA |
| NP\_004950.2 | c.1467G>A | rs151191539 | - | 427 | Arg>Gln | NA |
|  | c.1497T>A | rs104894120 VAR\_063258 | | 437 | Leu>Gln | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.190G>A | rs121918656 | - | 1 | Met>Ile | NA |
|  | c.230G>A | rs104894124 | VAR\_063255 | 15 | Val>Met | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.284T>A | - | VAR\_039106 | 33 | Cys>Ser | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.351G>A | rs184724257 | - | 55 | Cys>Tyr | NA |
|  | c.421G>A | rs104894125 | VAR\_063256 | 78 | Met>Ile | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.438G>A | - | VAR\_039107 | 84 | Arg>His | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.458G>A | rs104894126 VAR\_063257 | | 91 | Gly>Ser | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.462G>A | rs104894119 | VAR\_016982 | 92 | Arg>Gln | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.821G>A | - | VAR\_065868 | 212 | Gly>Ser | Spermatogenic failure type 8 (SPGF8) MIM:613957 |
|  | c.854G>A | rs74535046 | - | 223 | Val>Met | NA |
|  | c.899G>A | - | VAR\_065869 | 238 | Asp>Asn | Spermatogenic failure type 8 (SPGF8) MIM:613957 |
|  | c.956G>A | rs141502483 | - | 257 | Asp>Asn | NA |
|  | c.974G>A | rs143355429 | - | 263 | Gly>Ser | NA |
|  | c.285G>C | - | VAR\_039106 | 33 | Cys>Ser | 46 XY sex reversal type 3 (SRXY3) MIM:612965 |
|  | c.555G>C | - | VAR\_062967 | 123 | Gly>Ala | Premature ovarian failure type 7 (POF7) MIM:612964|Spermatogenic failure type 8 (SPGF8) MIM:61395 |
|  | c.624G>C | rs1110061 | VAR\_039108 | 146 | Gly>Ala | NA |
|  | c.1287A>G | rs76323457 | - | 367 | Glu>Gly | NA |
|  | c.198C>T | rs145936761 | - | 4 | Ser>Leu | NA |
|  | c.573C>T | - | VAR\_062968 | 129 | Pro>Leu | Premature ovarian failure type 7 (POF7) MIM:612964|Spermatogenic failure type 8 (SPGF8) MIM:61395 |
|  | c.579C>T | - | VAR\_065866 | 131 | Pro>Leu | Spermatogenic failure type 8 (SPGF8) MIM:613957 |
|  | c.758C>T | - | VAR\_065867 | 191 | Arg>Cys | Spermatogenic failure type 8 (SPGF8) MIM:613957 |
|  | c.951G>T | rs104894118 VAR\_016983 | | 255 | Arg>Leu | Adrenocortical insufficiency without ovarian defect (ACIWOD) MIM:184757 |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| GATA4 | c.1353G>A | rs116781972 | - | 267 | Val>Met | NA |
| NM\_002052.3 | c.1402G>A | rs180765750 | - | 283 | Arg>His | NA |
| NP\_002043.2 | c.1440G>A | rs104894073 VAR\_016204 | | 296 | Gly>Ser | Atrial septal defect type 2 (ASD2) MIM:607941 |
|  | c.1827G>A | rs56208331 | - | 425 | Asp>Asn | NA |
|  | c.1866G>A | - | - | 438 | Asp>Asn | NA |
|  | c.1174T>C | rs140892695 | - | 207 | Met>Thr | NA |
|  | c.1632G>C | rs141808522 | - | 360 | Glu>Gln | NA |
|  | c.1726G>C | rs147093296 | - | 391 | Gly>Ala | NA |
|  | c.609G>C | rs1139240 | - | 19 | Glu>Gln | NA |
|  | c.669G>C | rs1139241 | - | 39 | Val>Leu | NA |
|  | c.750G>C | rs1139244 | - | 66 | Ala>Pro | NA |
|  | c.832G>C | rs56191129 | - | 93 | Gly>Ala | NA |
|  | c.1500C>G | rs56298569 | - | 316 | Gln>Glu | NA |
|  | c.1682C>G | rs116414842 | - | 376 | His>Gln | NA |
|  | c.1683A>G | rs3729856 | VAR\_038196 | 377 | Ser>Gly | NA |
|  | c.1879C>G | rs146017816 | - | 442 | Ala>Gly | NA |
|  | c.1252C>T | - | - | 233 | Thr>Met | NA |
|  | c.1411C>T | rs116430078 | - | 286 | Ala>Val | NA |
|  | c.1660G>T | rs146685461 | - | 369 | Gly>Val | NA |
|  | c.1786C>T | rs55633527 | - | 411 | Ala>Val | NA |
|  | c.1866G>T | rs149351193 | - | 438 | Asp>Tyr | NA |
|  | c.709C>T | rs104894074 VAR\_038195 | | 52 | Ser>Phe | Atrial septal defect type 2 (ASD2) MIM:607941 |
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|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| MAMLD1 | c.1276C>A | rs62641609 | - | 322 | His>Gln | NA |
| NM\_001177465.1 | c.1289C>A | - | - | 327 | Pro>Thr | NA |
| NP\_001170936.1 | c.1439G>A | rs190798967 | - | 377 | Ala>Thr | NA |
|  | c.1791G>A | rs142352628 | - | 494 | Ser>Asn | NA |
|  | c.2103G>A | rs145175147 | - | 598 | Arg>His | NA |
|  | c.2996G>A | rs182200963 | - | 896 | Ala>Thr | NA |
|  | c.1749T>C | rs61740566 | - | 480 | Val>Ala | NA |
|  | c.1902A>C | rs113547218 | - | 531 | Lys>Thr | NA |
|  | c.3157A>C | rs148365369 | - | 949 | Glu>Asp | NA |
|  | c.981T>C | rs112342573 | - | 224 | Met>Thr | NA |
|  | c.982G>C | rs141528376 | - | 224 | Met>Ile | NA |
|  | c.1397C>G | - | - | 363 | Leu>Val | NA |
|  | c.1506A>G | rs148647178 | - | 399 | Gln>Arg | NA |
|  | c.1774C>G | rs61739630 | - | 488 | Asn>Lys | NA |
|  | c.1974A>G | - | VAR\_030025 | 555 | Gln>Arg | NA |
|  | c.686A>G | rs138334535 | - | 126 | Thr>Ala | NA |
|  | c.1221C>T | rs34108766 | - | 304 | Ser>Phe | NA |
|  | c.1256C>T | - | - | 316 | Arg>Cys | NA |
|  | c.1310C>T | rs41313406 | VAR\_030024 | 334 | Pro>Ser | NA |
|  | c.1362C>T | - | - | 351 | Ser>Leu | NA |
|  | c.1934C>T | rs146443503 | - | 542 | Pro>Ser | NA |
|  | c.2139C>T | rs140927620 | - | 610 | Ala>Val | NA |
|  | c.2319C>T | rs147056898 | - | 670 | Thr>Ile | NA |
|  | c.329C>T | - | - | 7 | Arg>Trp | NA |
|  | c.512C>T | rs145557326 | - | 68 | Leu>Phe | NA |
|  | c.552C>T | - | - | 81 | Ala>Val | NA |
|  | c.840C>T | - | - | 177 | Thr>Met | NA |
|  | c.975C>T | rs148317024 | - | 222 | Thr>Ile | NA |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| KAL1 | c.1301G>A | rs190088923 | - | 384 | Arg>Gln | NA |
| NM\_000216.2 | c.1360G>A | rs190618510 | - | 404 | Glu>Lys | NA |
| NP\_000207.2 | c.1682C>A | rs142729431 | - | 511 | Ser>Tyr | NA |
|  | c.1690G>A | rs137852515 VAR\_012742 | | 514 | Glu>Lys | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1701C>A | - | VAR\_031017 | 517 | Phe>Leu | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1739G>A | - | - | 530 | Ser>Asn | NA |
|  | c.1750G>A | rs808119 | VAR\_007721 | 534 | Val>Ile | NA |
|  | c.1765G>A | - | VAR\_065364 | 539 | Glu>Lys | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1777G>A | rs149299309 | - | 543 | Val>Ile | NA |
|  | c.1828G>A | rs2229013 | - | 560 | Val>Ile | NA |
|  | c.1861T>A | - | VAR\_031018 | 571 | Trp>Arg | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1904T>A | rs112190856 | - | 585 | Phe>Tyr | NA |
|  | c.2153G>A | - | VAR\_031020 | 668 | Arg>His | NA |
|  | c.586G>A | rs144620955 | - | 146 | Ala>Thr | NA |
|  | c.638G>A | - | VAR\_031012 | 163 | Cys>Tyr | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.748C>A | - | - | 200 | Leu>Met | NA |
|  | c.911G>A | - | - | 254 | Arg>Lys | NA |
|  | c.951T>A | - | VAR\_007720 | 267 | Asn>Lys | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1232T>C | - | - | 361 | Leu>Pro | NA |
|  | c.1699T>C | - | VAR\_031017 | 517 | Phe>Leu | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1861T>C | - | VAR\_031018 | 571 | Trp>Arg | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.533T>C | rs147475357 | - | 128 | Leu>Ser | NA |
|  | c.634A>C | - | - | 162 | Lys>Gln | NA |
|  | c.637T>C | - | VAR\_065363 | 163 | Cys>Arg | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.664T>C | - | VAR\_031013 | 172 | Cys>Arg | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.935G>C | - | VAR\_031014 | 262 | Arg>Pro | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1061A>G | - | VAR\_031015 | 304 | Asn>Ser | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1701C>G | - | VAR\_031017 | 517 | Phe>Leu | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.442A>G | - | - | 98 | Lys>Glu | NA |
|  |  |  |  |  |  |  |

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| --- | --- | --- | --- | --- | --- | --- |
|  | c.550T>G | - | VAR\_065362 | 134 | Cys>Gly | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.613A>G | rs143079588 | - | 155 | Asn>Asp | NA |
|  | c.951T>G | - | VAR\_007720 | 267 | Asn>Lys | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1337C>T | rs137852517 VAR\_031016 | | 396 | Ser>Leu | Kallmann syndrome type 1 (KAL1) MIM:308700 |
|  | c.1433C>T | rs148736113 | - | 428 | Pro>Leu | NA |
|  | c.1520G>T | rs147905733 | - | 457 | Arg>Leu | NA |
|  | c.1531C>T | rs144709908 | - | 461 | Arg>Trp | NA |
|  | c.1547C>T | rs192003597 | - | 466 | Ala>Val | NA |
|  | c.1909G>T | rs137900287 | - | 587 | Val>Leu | NA |
|  | c.2147A>T | - | VAR\_031019 | 666 | Lys>Met | NA |
|  | c.901A>T | - | - | 251 | Thr>Ser | NA |
|  |  |  |  |  |  |  |
| Gene and NCBI |  |  |  |  |  |  |
| Accession # | Coding nomenclature | dbSNP | UniProt | Protein position | wild-AA>Mutated AA | Disease |
| DMRT1 | c.1041T>A | rs147991046 | - | 298 | Ser>Thr | NA |
| NM\_021951.2 | c.1059G>A | rs141672484 | - | 304 | Val>Met | NA |
| NP\_068770.2 | c.153C>A | rs147366726 | - | 2 | Pro>Thr | NA |
|  | c.282T>A | rs3739583 | VAR\_009954 | 45 | Ser>Thr | NA |
|  | c.901C>A | rs150438352 | - | 251 | Ser>Tyr | NA |
|  | c.942G>A | rs146893441 | - | 265 | Val>Met | NA |
|  | c.990C>A | - | VAR\_009956 | 281 | Arg>Ser | NA |
|  | c.1140G>C | rs139434590 | - | 331 | Asp>His | NA |
|  | c.811A>C | - | VAR\_009955 | 221 | Tyr>Ser | NA |
|  | c.1018T>G | rs142683885 | - | 290 | Met>Arg | NA |
|  | c.1186A>G | rs35846503 | - | 346 | Lys>Arg | NA |
|  | c.673C>G | rs144989599 | - | 175 | Thr>Ser | NA |
|  | c.982T>G | rs79631603 | - | 278 | Met>Arg | NA |
|  | c.1033C>T | - | VAR\_009957 | 295 | Pro>Leu | NA |
|  | c.1111C>T | rs143383634 | - | 321 | Ala>Val | NA |
|  | c.1187G>T | rs146782823 | - | 346 | Lys>Asn | NA |
|  | c.177C>T | rs139583942 | - | 10 | Pro>Ser | NA |
|  | c.921C>T | rs188566489 | - | 258 | Arg>Trp | NA |

# Supplemental Table 1: All the DSD genes with their genetic variations were listed as mined from MutDB <http://www.mutdb.org/>, with dbSNP <https://www.ncbi.nlm.nih.gov/snp> and UniProt Ids <https://www.uniprot.org/> as well as disease phenotype from OMIM <http://omim.org/> .

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