**The DSD panel contains genes involved in gonadal development and gonadal differentiation (PanelApp v2.1, January 2020)**

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| **Disorders of gonadal development** | | |
| **Gene** | **Associated phenotypes** | **OMIM** |
| *ARX* | Proud syndrome (Corpus Callosum, Agenesis Of, With Abnormal Genitalia)  X-linked Lissencephaly type 2 | [300004](https://www.omim.org/entry/300004)  [300215](https://www.omim.org/entry/300215) |
| *ATRX* | X-linked Alpha-thalassemia/mental retardation syndrome | [301040](https://www.omim.org/entry/301040) |
| *CUL4B* | X-linked intellectual disability, Cabezas type | [300354](https://omim.org/entry/300354) |
| *DHH* | 46,XY complete gonadal dysgenesis (46,XY sex reversal type 7)  46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome | [233420](https://omim.org/entry/607080)  [607080](https://omim.org/entry/607080) |
| *MAMLD1* | X-linked Hypospadias type 2  X-linked myotubular myopathy-abnormal genitalia syndrome | [300758](https://omim.org/entry/300120?search=mamld1&highlight=mamld1)  [300219](https://omim.org/entry/300219) |
| *MAP3K1* | 46,XY complete gonadal dysgenesis (46,XY sex reversal type 6) 46,XY partial gonadal dysgenesis | [613762](https://www.omim.org/entry/613762) |
| *NR0B1* | 46,XY sex reversal type 2  Congenital Adrenal Hypoplasia | [300018](https://www.omim.org/entry/300018)  [300200](https://www.omim.org/entry/300200) |
| *NR5A1* | 46,XX sex reversal type 4  46,XY sex reversal type 3  Adrenocortical insufficiency/Premature Ovarian Failure type 7  Spermatogenic failure type 8 | [617480](https://www.omim.org/entry/617480)  [612965](https://www.omim.org/entry/612965)  [612964](https://www.omim.org/entry/612964)  [613957](https://www.omim.org/entry/613957) |
| *RPL10* | Syndromic X-linked intellectual disability | [300998](https://www.omim.org/clinicalSynopsis/300998) |
| *RSPO1* | Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal | [610644](https://www.omim.org/entry/610644) |
| *SOX9* | Campomelic dysplasia | [114290](https://www.omim.org/entry/114290) |
| *SOX10* | Peripheral demyelinating neuropathy, Central dysmyelination, Waardenburg syndrome, and Hirschsprung disease (PCWH)  Waardenburg syndrome, type 4C | [609136](https://www.omim.org/entry/609136)  [613266](https://www.omim.org/entry/613266) |
| *SRY* | 46,XX sex reversal type 1  46,XY sex reversal type 1 | [400045](https://www.omim.org/entry/400045) [400044](https://www.omim.org/entry/400044) |
| *TOE1* | Pontocerebellar hypoplasia, type 7 | [614969](https://www.omim.org/entry/614969) |
| *WT1* | Denys-Drash syndrome  Frasier syndrome | [194080](https://www.omim.org/entry/194080)  [136680](https://www.omim.org/entry/136680) |
| **Disorders of gonadal differentiation** | | |
| **Gene** | **Associated phenotypes** | **OMIM** |
| *AMH* | Persistent Mullerian duct syndrome type 1 | [261550](https://www.omim.org/entry/261550) |
| *AMHR2* | Persistent Mullerian duct syndrome type 2 | [261550](https://www.omim.org/entry/261550) |
| *AR* | Androgen insensitivity  X-linked Hypospadias type 1 | [300068](https://www.omim.org/entry/300068)  [300633](https://www.omim.org/entry/300633) |
| *CDKN1C* | IMAGE syndrome | [614732](https://www.omim.org/entry/614732) |
| *CHD7* | CHARGE syndrome  Hypogonadotropic hypogonadism type 5 | [214800](https://www.omim.org/entry/214800)  612370 |
| *CYB5A* | Methemoglobinemia and ambiguous genitalia | [250790](https://www.omim.org/entry/250790) |
| *CYP11A1* | Congenital adrenal insufficiency with 46,XY sex reversal | [613743](https://www.omim.org/entry/613743) |
| *CYP11B1* | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency  Glucocorticoid-remediable aldosteronism | [202010](https://www.omim.org/entry/202010)  [103900](https://www.omim.org/entry/103900) |
| *CYP17A1* | 46,XY disorder of sex development due to isolated 17,20-lyase deficiency | [202110](https://omim.org/entry/202110) |
| *CYP19A1* | Aromatase excess syndrome | [139300](https://omim.org/entry/139300) |
| *CYP21A2\** | Congenital adrenal hyperplasia due to 21-hydroxylase deficiency | [201910](https://omim.org/entry/201910) |
| *DHCR7* | Smith-Lemli-Opitz syndrome | [270400](https://omim.org/entry/270400) |
| *HSD17B3* | 46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency | [264300](https://omim.org/entry/264300) |
| *HSD3B2* | Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency | [201810](https://omim.org/entry/201810) |
| *LHCGR* | Leydig cell hypoplasia due to complete LH resistance | [238320](https://omim.org/entry/238320) |
| *POR* | Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis | [201750](https://omim.org/entry/201750) |
| *SAMD9* | MIRAGE syndrome | [617053](https://omim.org/entry/617053) |
| *SGPL1* | Nephrotic syndrome type 14 | [617575](https://omim.org/entry/617575) |
| *SRD5A2* | 46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency | [264600](https://omim.org/entry/264600) |
| *STAR* | Lipoid adrenal hyperplasia | [201710](https://omim.org/entry/201710) |

\*Due to the presence of pseudogenes with very high sequence similarity, mapping of reads over this gene is suboptimal. If a diagnosis of Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency (CAH) is suspected please request additional testing (Sanger sequencing and copy number analysis by MLPA).