Additional file 2: Table S1: OMIM genes in the overlapping deleted region on 10q22.2

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| --- | --- | --- | --- | --- | --- |
| OMIM ID | Gene | | Associated Disorder | Inheritance | Clinical Presentation |
| Symbol | Title |
| 605880 | KAT6B | Lysin Acetyltransferase 6B (Histone acetyltransferase) | SBBYSS syndrome | Autosomal Dominant | Major Features: Long thumbs/great toes, immobile mask-like face (expressionless face), blepharophimosis/ptosis, lacrimal duct anomalies, and patellar hypoplasia/agenesis Minor Features: Congenital heart defect, dental anomalies, hearing loss, thyroid anomalies, anal anomalies, hypotonia, and global developmental delay/intellectual disability |
| Genitopatellar syndrome | Autosomal Dominant | Major Features: Genital anomalies, Patellar hypoplasia/agenesis, flexion contractures at the hips and knees, club feet, agenesis of the corpus callosum, microcephaly, hydronephrosis and/or multiple renal cysts Minor Features: Congenital heart defect, Dental anomalies, hearing loss, thyroid anomalies, anal anomalies, hypotonia, and global developmental delay/intellectual disability |
| 613191 | DUSP13 | Dual-specificity phosphatases 13 | None | NA | NA |
| 611575 | SAMD8 | Sterile Alpha Motif Domain-containing Protein 8 | None | NA | NA |
| 193245 | VDAC2 | Voltage-Dependent Anion Channel 2 | None | NA | NA |
| 613902 | ZNF503 | Zinc Finger Protein 503 | None | NA | NA |
| 614537 | C10ORF11 | Chromosome 10 Open Reading Frame 11 | Albinism, oculocutaneous | Autosomal recessive | Abnormal pigmentation, photophobia, and nystagmus |

NA- Not applicable