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| --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient ID** | **Karyotype** | **Start and end breakpoints of 16q deletion (GRCh37/hg19)** | **Size (Mb)** | **Classification** | **Inheritance** | **Further CNVs** | **Affected genes** |
| DECIPHER 402414 | 46,XX | 74,639,048-78,901,055 | 4.262 | LP | Maternal | No | *ADAMTS18, CHST6, FA2H, KARS, LDHD, RFWD3, TMEM231, WWOX* |
| DECIPHER 297860 | 46,XY | 76,180,903-82,314,260 | 6.133 | P | De novo | 1 VUS | *ADAMTS18, BCMO1, GAN, GCSH, MAF, PLCG2, WWOX* |
| DECIPHER 306151 | 46,XX | 76,849,848-83,159,043 | 6.309 | P | unknown | 1 VUS | *ADAMTS18, BCMO1, GAN, GCSH, MAF, PLCG2, WWOX* |
| nssv577564 | n.a. | 69,951,979-76,757,245 | 6.805 | P | n.a. | n.a. | *AARS, C16orf47, CHST6, COG4, DHODH, DHX38, FA2H, FUK, HP, HYDIN, KARS, LDHD, MIR140, NQO1, PMFBP1, RFWD3, TAT, TMEM231, VAC14, ZFHX3* |
| DECIPHER 295733 | 46,XX | 78,738,853-85,707,367 | 6.939 | VUS | De novo | No | *BCMO1, DNAAF1, GAN, GCSH, MAF, MBTPS1, MLYCD, PLCG2, SLC38A8, WWOX* |
| DECIPHER 285735 | 46,XY | 72,125,903-80,637,770 | 8.512  | P | De novo | No | *ADAMTS18, C16orf47, CHST6, DHX38, FA2H, KARS, LDHD, MAF, PMFBP1, RFWD3, TMEM231, WWOX, ZFHX3* |
| DECIPHER 396469 | 46,XY | 74,142,499-83,242,499 | 9.100 | LP | De novo | No | *ADAMTS18, BCMO1, CHST6, FA2H, GAN, GCSH, KARS, LDHD, MAF, PLCG2, RFWD3, TMEM231, WWOX* |
| nssv577565 | n.a. | 73,083,366-82,609,931 | 9.567 | P | n.a. | n.a. | *ADAMTS18, BCMO1, C16orf47, CHST6, FA2H, GAN, GCSH, KARS, LDHD, MAF, PLCG2, RFWD3, TMEM231, WWOX, ZFHX3* |
| **DECIPHER414204 (Proband)** | **46,XY** | **72,155,844-82,148,404** | **9.993** | **LP** | **De novo** | **No** | ***ADAMTS18, BCMO1, C16orf47, CHST6, FA2H, GAN, GCSH, KARS, LDHD, MAF, PLCG2, PMFBP1, RFWD3, TMEM231, WWOX, ZFHX3*** |
| DECIPHER 395155 | 46,XX | 66,542,499-76,542,499 | 10.000 | LP | De novo | No | *AARS, ACD, AGRP, C16orf47, CBFB, CDH1, CDH3, CENPT, CHST6, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, NOL3, NQO1, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, VAC14, ZFHX3* |
| DECIPHER 395609 | 46,XX | 76,542,499-89,972,499 | 13.430 | LP | De novo | No | *ACSF3, ADAMTS18, ANKRD11, APRT, BCMO1, CA5A, CDH15, CDK10, CDT1, CHMP1A, CTU2, CYBA, DNAAF1, FANCA, FBXO31, FOXC2, FOXF1, GALNS, GAN, GCSH, IRF8, JPH3, MAF, MBTPS1, MLYCD, MVD, PIEZO1, PLCG2, RPL13, SLC38A8, SPG7, TRAPPC2L, WWOX, ZNF469* |
| DECIPHER 396509 | 46,XY | 76,542,499-89,972,499 | 13.430 | LP | De novo | No | *ACSF3, ADAMTS18, ANKRD11, APRT, BCMO1, CA5A, CDH15, CDK10, CDT1, CHMP1A, CTU2, CYBA, DNAAF1, FANCA, FBXO31, FOXC2, FOXF1, GALNS, GAN, GCSH, IRF8, JPH3, MAF, MBTPS1, MLYCD, MVD, PIEZO1, PLCG2, RPL13, SLC38A8, SPG7, TRAPPC2L, WWOX, ZNF469* |
| nssv576729 | n.a. | 70,448,476-84,941,726 | 14.493 | P | n.a. | n.a. | *AARS, ADAMTS18, BCMO1, C16orf47, CHST6, COG4, DHODH, DHX38, DNAAF1, FA2H, FUK, GAN, GCSH, HP, HYDIN, KARS, LDHD, MAF, MBTPS1, MLYCD, PLCG2, PMFBP1, RFWD3, SLC38A8, TAT, TMEM231, VAC14, WWOX, ZFHX3* |
| DECIPHER 395170 | 46,XX | 66,542,499-81,242,499 | 14.700 | LP | De novo | No | *AARS, ACD, ADAMTS18, AGRP, C16orf47, CBFB, CDH1, CDH3, CENPT, CHST6, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, GCSH, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, MAF, NOL3, NQO1, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, VAC14, WWOX, ZFHX3* |
| DECIPHER 288191 | 46,XY | 75,036,313-85,730,707 | 16.694 | VUS | De novo | 1 VUS | *ADAMTS18, BCMO1, CHST6, DNAAF1, GAN, GCSH, KARS, LDHD, MAF, MBTPS1, MLYCD, PLCG2, SLC38A8, TMEM231, WWOX* |
| DECIPHER 395094 | 46,XX | 66,542,499-83,242,499 | 16.700 Mb | LP | De novo | No | *AARS, ACD, ADAMTS18, AGRP, BCMO1, C16orf47, CBFB, CDH1, CDH3, CENPT, CHST6, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, GAN, GCSH, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, MAF, NOL3, NQO1, PLCG2, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, VAC14, WWOX, ZFHX3* |
| DECIPHER 395079 | 46,XX | 57,942,499-76,542,499 | 18.600 | LP | De novo | No | *AARS, ACD, AGRP, BEAN1, C16orf47, CBFB, CDH1, CDH11, CDH3, CENPT, CHST6, CNGB1, CNOT1, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, GOT2, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, NOL3, NQO1, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, USB1, VAC14, ZFHX3* |
| DECIPHER 393303 | 46,XX | 57,942,499-76,542,499 | 18.600 | LP | De novo | No | *AARS, ACD, AGRP, BEAN1, C16orf47, CBFB, CDH1, CDH11, CDH3, CENPT, CHST6, CNGB1, CNOT1, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, GOT2, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, NOL3, NQO1, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, USB1, VAC14, ZFHX3* |
| DECIPHER 393344 | 46,XY | 57,942,499-76,542,499 | 18.600 | LP | De novo | No | *AARS, ACD, AGRP, BEAN1, C16orf47, CBFB, CDH1, CDH11, CDH3, CENPT, CHST6, CNGB1, CNOT1, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, GOT2, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, NOL3, NQO1, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, USB1, VAC14, ZFHX3* |
| DECIPHER 394950 | 46,XY | 57,942,499-76,542,499 | 18.600 | LP | De novo | No | *AARS, ACD, AGRP, BEAN1, C16orf47, CBFB, CDH1, CDH11, CDH3, CENPT, CHST6, CNGB1, CNOT1, COG4, COG8, CTCF, DHODH, DHX38, FA2H, FUK, GOT2, HP, HSD11B2, HSF4, HYDIN, KARS, LCAT, LDHD, NOL3, NQO1, PMFBP1, PRMT7, RFWD3, RLTPR, TAT, TK2, TMEM231, USB1, VAC14, ZFHX3* |

**Supplementary Table 2. Genotype comparison of the index patient and overlapping cases from public databases.**
Abbreviations: Mb: megabase; VUS: variant of unknown significance; LP: likely pathogenic; P: pathogenic; CNV: copy number variant; DD: developmental delay; ID: intellectual disability; n.a.: not available