

Supplementary Table SII Details of HGNC Genes identified within CNVs in 46, XY DSD boys. Information was not available for cases 72 and 121.

| Subject Ref | CNV | Protein coding genes | Non-protein coding genes |
|-------------|--------------|--|--|
| 24 | Del 7q34 | <i>MTRNR2L6, PRSS1</i> | <i>PGBD4P1, TRBV10-1, TRBV11-1, TRBV12-1, TRBV19, TRBV20-1, TRBV21-1, TRBV22-1, TRBV23-1, TRBV24-1, TRBV25-2, TRBV26, TRBV27, TRBV28, TRBV29-1, TRBV5-2, TRBV5-3, TRBV6-4, TRBV7-3, TRBV8-1, TRBV8-2, TRBV9, TRBVA, TRBVB</i> |
| 28 | Del 2p.22.3 | Does not span any genes | Does not span any genes |
| 40 | Del 4q13.3 | <i>ADAMTS3, NPFFR2</i> | <i>HNRNPA1P67</i> |
| | Del 16p12.2 | <i>IGSF6, METTL9, NPIP4, OTOA,</i> | <i>RNU6-1005P, RNU6-196P, RRN3P1</i> |
| | Dup 20p12.3 | <i>PLCB1, PLCB4</i> | |
| 56 | Del 1q31.1 | Does not span any genes | Does not span any genes |
| | Del 5p14.3 | <i>CDH18</i> | <i>GUSBP1</i> |
| | Dup 13q32.1 | <i>FARP1, HS6ST3, IPO5, MBNL2, OXGR1, RAP2A, RNF113B, STK24</i> | <i>FARP1-AS1, FTLP8, HSP90AB6P, LINC00359, LINC00456, MIR3170, PSMA6P4, RN7SKP7, RN7SKP8, RNA5SP37, RPL7AP61, TULP3P1</i> |
| 65 | Del 12q13.12 | <i>AQP2, AQP5, AQP6, BCDIN3D, FAIM2, FAM186B, FMNL3, KCNH3, MCRS1, NCKAP5L, PRPF40B, RACGAP1, SPATS2, TMBIM6,</i> | <i>BCDIN3D-AS1, HIGD1AP9, LSM6P2, POLR2KP1, RNU6-834P</i> |
| 69 | Dup 15q11.1 | <i>CYFIP1, GOLGA6L1, GOLGA6L2, GOLGA6L6, MKRN3, NIPA1, NIPA2, OR4M2, OR4N4, POTE1, POTE2, TUBGCP5</i> | <i>ABCB10P1, BMS1P16, CT60, CXADRP2, ELMO2P1, GOLGA8CP, GOLGA8DP, GOLGA8EP, GOLGA8I, GOLGA8S, GRAMD4P5, GRAMD4P6, HERC2P2, HERC2P3, HERC2P7, IGHDIOR25-1B, IGHDIOR25-2B, IGHDIOR25-3B, IGHDIOR25-4B, IGHDIOR25-5B, IGHDIOR25-6, IGHDIOR25-7, IGHDIOR25-8, KIAA0125P2, MIR1268A, MIR3118-4, MIR3118-6, MIR4508, MIR4509-1, MIR5701-1, MIR5701-2, NBEAP1, ORI1H3P, ORI1J1P, ORI1J2P, ORI1J5P, ORI1K1P, OR4H6P, OR4N3P, OR4Q1P, RN7SL106P, RN7SL400P, RN7SL495P, RN7SL536P, RN7SL545P, RN7SL759P, RNU6-1235P, RNU6-498P, RNU6-631P, RNU6-749P, SPATA31E2P, SPATA31E3P, TUBGCP5, WHAMMP3</i> |
| 82 | Dup 16p11.2 | Does not span any genes | Does not span any genes |
| 86 | Dup 2p16.3 | <i>FSHR, GTF2A1L, LHCGR, STON1, STON1-GTF1L, TCEB2P3, TPT1P11</i> | <i>CTBP2P5, RNU6-439P, STON1-GTF1L, TCEB2P3, TPT1P11</i> |
| 91 | Dep 18q21.32 | <i>ADNP2, ALPK2, ATP9B, BCL2, C18orf63, CBLN2, CCBE1, CCDC102B, CD226, CDH19, CDH20, CDH7, CNDP1, CNDP2, CPLX4, CTDP1, CYB5A, DOK6, DSEL, FAM69C, FBXO15, GALR1, GRP, HMSD, HSBP1L1, KCNG2, KDSR, KIAA1468, LMAN1, MALTI, MBP, MC4R, NETO1, NFATC1, PARD6G, PHLPP1, PIGN, PMAIP1, PQLC1, RAX, RBFA, RNF152, RTTN, SALL3, SEC11C, SERPINB11, SERPINB12, SERPINB13, SERPINB1, SERPINB2, SERPINB3, SERPINB4, SERPINB5, SERPINB7, SERPINB8, SIMIM21, SOCS6, TIMM21, TMX3, TNFRSF11A, TSHZ1, TXNL4A, VPS4B, ZADH2, ZCCHC2, ZNF236, ZNF407, ZNF516, ZNF532</i> | <i>ACTBP9, AKR1B10P2, ARL2BPP1, ATP5G1P6, ATP9B, BDP1P, CTBP2P3, FAM60CP, FAU1, GLUD1P4, GTSCR1, HNRNPA1P11, LINC00305, LINC00683, LINC00908, LINC00909, LINC01029, MIR5011, MIR548AV, MRPL37P1, MRPS5P4, NFE2L3P1, OACYLP, PRPF19P1, RBFADN, RN7SL112P, RN7SL342P, RN7SL401P, RN7SL551P, RN7SL705P, RN7SL795P, RNA5SP461, RNU2-69P, RNU4-17P, RNU6-1037P, RNU6-116P, RNU6-142P, RNU6-219P, RNU6-346P, RNU6-39P, RNU6-567P, RNU6-655P, RNU7-146P, RPIAP1, RPL12P39, RPL17P44, RPL26P35, RPL30P14, RPL31P9, RPL9P31, RPS2P6, RPS3AP49, SDCCAG3P1, SDHCP1, SERPINB10, SLC25A6P4,</i> |
| 99 | Del 20p13 | <i>ANGPT4, C20orf202, C20orf96, CSNK2A1, DEFB125, DEFB126, DEFB127, DEFB128, DEFB129, DEFB132, FAM110A, FKBP1A, NRSN2, NSFL1C, PSMF1, RAD21L1, RBCK1, RSPO4, SCRT2, SCRT2, SDCBP2, SIRPB1, SIRPB2, SIRPD, SIRPG, SLC52A3, SNPH, SOX12, SRXN1, TBC1D20, TCF15, TMMEM74B, TRIB3, ZCCHC3,</i> | <i>ACTG1P3, RN7SL561P, RNU6-917P, SDCBP2-AS1, SIRPB3P,</i> |
| 132 | Dup 11q11q | <i>OR10AG1, OR4A15, OR4A16, OR4C11, OR4C15, OR4C16, OR4C6, OR4P4, OR4S2, OR5AS1, OR5D13, OR5D14, OR5D16, OR5D18, OR5F1, OR5I1, OR5J2, OR5L1, OR5L2, OR5W2, OR8H2, OR8K3, OR8J2, OR8J3, OR8K5, TRIM51</i> | <i>OR10AF1P, OR10AK1P, OR4A10P, OR4A11P, OR4A13P, OR4A14P, OR4A17P, OR4A21P, OR4A50P, OR4A9P, OR4C11, OR4C14P, OR4P1P, OR4VIP, OR4X7P, OR5AQ10, OR5BE1P, OR5BN1P, OR5BN2P, OR5D17P, OR5D2P, OR5D3, OR5F2P, OR5J1P, OR5J7P, OR5W1P, OR7E5P, OR8H3, OR8I4P, OR8K4P, OR8V1P, OR9M1P</i> |

Abbreviations: HGNC, HUGO Gene Nomenclature Committee; CNV, copy number variants; Dup, duplication; Del, deletion.