**Supplemental Table I. TaqMan copy number assay details**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Locus | Assay ID | Gene | Target Coordinates | # Cases Tested | # Controls Tested |
| 1p35.1 | Hs04545441\_cn | ADC | Chr.1:33,551,372 | 57 | 38 |
| 1p35.1 | Hs01582102\_cn | TRIM62 | Chr.1:33,623,863 | 57 | 181 |
| 1p34.1 | Hs03380682\_cn | PIK3R3 | Chr.1:46,598,687 | Probe Excluded |
| 1p34.1 | Hs05796956\_cn | - | Chr.1:46,637,647 | 57 | 181 |
| 1p31.1 | Hs05798253\_cn | - | Chr.1:82,957,446 | 57 | 181 |
| 1p31.1 | Hs05737448\_cn | - | Chr.1:83,074,614 | 57 | 32 |
| 1q21.3 | Hs01440205\_cn | LCE1B | Chr.1:152,785,406 | 57 | 32 |
| 1q21.3 | Hs05796579\_cn | - | Chr.1:153,102,966 | 57 | 33 |
| 1q21.3 | Hs04525355\_cn | - | Chr.1:153,207,602 | 57 | 181 |
| 2p15 | Hs04662768\_cn | - | Chr.2:63,961,800 | 57 | 181 |
| 2p15 | Hs04692212\_cn | - | Chr.2:64,053,511 | 57 | 32 |
| 2p15 | Hs04696567\_cn | UGP2 | Chr.2:64,090,441 | 57 | 32 |
| 3p25.3 | Hs05903486\_cn | IRAK2 | Chr.3:10,259,290 | 57 | 32 |
| 3p25.3 | Hs00933380\_cn | GHRL | Chr.3:10,327,453 | 57 | 181 |
| 3q23 | Hs02313216\_cn | RBP2 | Chr.3:139,171,955 | 57 | 32 |
| 3q23 | Hs05901157\_cn | - | Chr.3:139,525,564 | 57 | 181 |
| 3q23 | Hs03487736\_cn | CLSTN2 | Chr.3:140,209,817 | 57 | 32 |
| 3q28 | Hs06618774\_cn | FGF12 | Chr.3:191,909,883 | 57 | 181 |
| 3q28 | Hs05888061\_cn | FGF12 | Chr.3:191,931,604 | 57 | 32 |
| 4q35.2 | Hs04880782\_cn | - | Chr.4:187,452,339 | 57 | 36 |
| 4q35.2 | Hs04873195\_cn | FAT1 | Chr.4:187,558,970 | 57 | 181 |
| 6p22.3 | Hs06778434\_cn | ATXN1 | Chr.6:16,451,082 | 57 | 181 |
| 6p22.3 | Hs06742415\_cn | - | Chr.6:16,841,101 | 57 | 32 |
| 6q24.1 | Hs04915424\_cn | - | Chr.6:140,785,204 | 57 | 181 |
| 6q24.1 | Hs06822965\_cn | - | Chr.6:141,127,772 | 57 | 32 |
| 6q24.1 | Hs06806879\_cn | - | Chr.6:142,168,485 | 57 | 32 |
| 12p13.2 | Hs06933845\_cn | - | Chr.12:10,879,713 | 57 | 181 |
| 12p13.2 | Hs06355595\_cn | PRH1-PRR4 | Chr.12:11,027,653 | 57 | 32 |
| 16p13.12 | Hs05413230\_cn | - | Chr.16:13,948,596 | 57 | 32 |
| 16p13.12 | Hs05403971\_cn | - | Chr.16:14,361,457 | 57 | 181 |
| 17q23.1 | Hs03959616\_cn | HEATR6 | Chr.17:58,122,375 | 57 | 32 |
| 17q23.2 | Hs05521790\_cn | TBX4 | Chr.17:59,546,983 | 57 | 32 |
| 17q23.2 | Hs01156556\_cn | MED13 | Chr.17:60,022,358 | 57 | 181 |
| 18q21.31 | Hs06494556\_cn | NEDD4L | Chr.18:56,022,110 | 57 | 33 |
| 18q21.31 | Hs06482541\_cn | - | Chr.18:56,104,534 | 57 | 181 |
| 20q13.2 | Hs02620199\_cn | PFDN4 | Chr.20:52,824,508 | 57 | 181 |
| 20q13.31 | Hs07180535\_cn | BMP7 | Chr.20:55,832,249 | 57 | 34 |
| 20q13.31 | Hs04037960\_cn | PMEPA1 | Chr.20:56,265,886 | 57 | 32 |
| 21q22.11 | Hs05551757\_cn | - | Chr.21:31,526,506 | 57 | 35 |
| 21q22.11 | Hs00141231\_cn | CLDN17 | Chr.21:31,538,308 | 57 | 181 |

**Supplemental Table II.** Additionalrare CNVs identified but not followed up in patients with isolatedposterior urethral valves

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Locus** | **Genomic Coordinates** | **Size (bps)** | **Type** | **Study Subject****ID** | **DECIPHER patients with overlapping CNVs and kidney and urinary tract anomalies** | **Gene(s)/Transcript(s)** |
| 1p22.3 | 87,017,093..87,087,981 | 70,889 | Dupl | 25 | 266358: Hydronephrosis | *AL356270.1; CLCA4; RP4-651E10.4* |
| 2p16.1 | 57,062,100..57,237,043 | 174,944 | Het Del | 26 |  | intergenic |
| 2p24.1 | 21,822,194..21,859,476 | 37,283 | Het Del | 22 | 282138: Micropenis | *AC009411.2; AC067959.1* |
| 2q21.2 | 134,635,194..134,657,542 | 22,349 | Het Del | 19 | 251953: abnormality of the kidney | intergenic |
| 2q31.1 | 172,353,544..172,460,954 | 107,411 | Dupl | 16 | 258540: Micropenis,275286: Structural anomalies of the renal tract | *CYBRD1* |
| 2q36.3 | 229,153,079..229,190,234 | 37,156 | Het Del | 5 |  | intergenic |
| 3p25.1 | 14,590,287..14,610,761 | 20,475 | Dupl | 25 |  | intergenic |
| 3q26.1 | 165,942,772..165,988,394 | 45,623 | Dupl | 27 | 250665: Renal hypoplasia | intergenic |
| 4q22.1 | 92,186,516..92,253,477 | 66,962 | Dupl | 18 | 753: Decreased testicular size | *FAM190A; RP11-763F8.1* |
| 4q24 | 107,110,343..107,180,404 | 70,062 | Dupl | 23 |  | *TBCK* |
| 4q25 | 110,019,097..110,152,741 | 133,645 | Het Del | 15 |  | *COL25A1* |
| 4q28.2 | 130,783,282..130,803,928 | 20,647 | Dupl | 31 |  | *RP11-422J15.1* |
| 4q28.2 | 130,983,055..131,048,431 | 65,377 | Het Del | 20 |  | intergenic |
| 5q21.1\* | 98,776,240..98,906,525 | 130,286 | Dupl | 17 |  | *CTD-2151A2.1* |
| 5q34 | 160,251,449..160,275,493 | 24,045 | Het Del | 29 |  | *ATP10B* |
| 8p22\* | 17,614,003.. 17,709,021 | 95,019 | Dupl | 17 | 253970: Abnormality of the kidney267866: Micropenis | *MTUS1; RP11-156K13.1;**RP11-806O11.1* |
| 8q21.3 | 81,146,729..81,166,822 | 20,094 | Het Del | 21 |  | intergenic |
| 11q23.1 | 110,989,288..111,129,765 | 140,478 | Dupl | 23 | 258577: Micropenis | intergenic |
| 11q14.3 | 89,875,437.. 90,091,334 | 215,898 | Dupl | 3 |  | *AP002364.1; CHORDC1; NAALAD2; RP11-660M18.2* |
| 13q31.3 | 92,061,878..92,302,332 | 240,455 | Het Del | 1 | 270910: Abnormality of the genital system | *GPC5* |
| 15q21.3 | 55,567,881..55,710,369 | 142,489 | Dupl | 28 | 260222: Micropenis | *CCPG1; DYX1C1-CCPG1; MIR628; PIGB; RAB27A; RP11-139H15.1; RP11-178D12.1* |
| 15q21.3 | 55,798,599..55,941,624 | 143,026 | Dupl | 28 | 260222: Micropenis | *DYX1C1-CCPG1; PRTG; PYGO1* |
| 16p11.2 | 31,376,849..31,402,514 | 25,666 | Dupl | 24 | 264947: Hypospadias | *ITGAX* |
| 16p13.13 | 10,594,596..10,618,314 | 23,719 | Het Del | 19 | 264947: Hypospadias | *RP11-27M24.3* |
| 16q22.2 | 72,694,086..72,764,017 | 69,932 | Het Del | 30 |  | *AC004158.2; RP5-991G20.1; U7* |
| 20q12 | 41,550,552..41,631,376 | 80,825 | Het Del | 22 |  | *PTPRT;RP4-753D4.2* |
| Xq21.32 | 92,312,127..92,358,210 | 46,084 | Het Del\*\* | 10 |  | intergenic |
| Xq23 | 111,665,007..111,796,169 | 131,163 | Dupl | 32 | 249479: Hypospadias | *ZCCHC16* |
| Xq28 | 148,686,062..148,736,128 | 50,067 | Dupl | 5 |  | *TMEM185A* |
| Xq28 | 153,184,816..153,579,448 | 394,633 | Dupl | 22 | 4571: Micropenis, 267999: Scrotal hypoplasia, 286121: Hypospadias and Micropenis | *ARHGAP4*; *NAA10*; *RENBP*; *HCFC1*; *TMEM187*; *MIR3202-1*; *TMEM187*; *IRAK1*; *MIR718*; *MECP2*; *OPN1LW*; *TEX28*; *OPN1MW*; *TKTL1*; *FLNA* |

Abbreviations: Het Del: Heterozygous deletion, Dupl: Duplication.

Coordinates (hg19) predicted using PennCNV.

\*Originally called as two separate CNVs, PennCNV’s clean\_cnv.pl function merged them into 1 with the reported coordinates.

\*\* The chromosome X call at 92,312,127-92,358,210 is at the 3’ end of a pseudoautosomal region (PAR3). Males have two copies of the genes in these regions, one on the X chromosome and one on the Y chromosome. This male individual appears to have only one copy of this section of PAR3, and it is not possible to determine from the array data whether this region has been deleted from the X or Y chromosome.