

Supplemental Table SI. TaqMan copy number assay details

| Locus | Assay ID | Gene | Target Coordinates | # Cases Tested | # Controls Tested |
|---------|---------------|-----------------|--------------------|----------------|-------------------|
| 1p34.2 | Hs03369968_cn | <i>CCDC30</i> | Chr.1:43,018,088 | 34 | 198 |
| | Hs03355957_cn | | Chr.1:43,031,517 | 34 | 24 |
| 1q25.3 | Hs00499239_cn | <i>COLGALT2</i> | Chr.1:183,923,980 | 34 | 25 |
| | Hs05762474_cn | - | Chr.1:184,184,766 | 34 | 198 |
| 2p12 | Hs05813312_cn | - | Chr.2:81,206,766 | 34 | 198 |
| | Hs04677525_cn | | Chr.2:81,331,795 | 34 | 24 |
| 3q26.32 | Hs04741060_cn | - | Chr.3:176,979,148 | 34 | 24 |
| | Hs06667233_cn | | Chr.3:176,987,683 | 34 | 198 |
| 4q22.3 | Hs04852209_cn | <i>BMPRI3</i> | Chr.4:95,987,323 | 34 | 24 |
| | Hs02817570_cn | | Chr.4:96,025,606 | 34 | 24 |
| | Hs04866592_cn | <i>UNC5C</i> | Chr.4:96,112,131 | 34 | 198 |
| 4q24 | Hs05939319_cn | - | Chr.4:104,837,852 | 34 | 24 |
| | Hs04866497_cn | | Chr.4:104,982,988 | 34 | 198 |
| | Hs05932226_cn | | Chr.4:105,031,088 | 34 | 24 |
| 4q28.3 | Hs05912262_cn | - | Chr.4:133,019,199 | 34 | 24 |
| | Hs04849484_cn | | Chr.4:133,037,687 | 34 | 198 |
| | Hs04883161_cn | | Chr.4:133,028,670 | 34 | 24 |
| 5p15.2 | Hs06018549_cn | <i>CTNND2</i> | Chr.5:11,868,014 | 34 | 198 |
| | Hs05998078_cn | | Chr.5:11,880,196 | 34 | 25 |
| 6p24.3 | Hs04914073_cn | <i>GCNT2</i> | Chr.6:10,533,075 | 34 | 25 |
| | Hs03618929_cn | <i>C6orf52</i> | Chr.6:10,684,894 | 34 | 198 |
| 6p12.3 | Hs06161050_cn | - | Chr.6:49,785,779 | 34 | 24 |
| | Hs06153060_cn | <i>CRISPI</i> | Chr.6:49,804,497 | 34 | 198 |
| 6q12 | Hs03089586_cn | - | Chr.6:66,978,264 | 34 | 198 |
| | Hs06795352_cn | | Chr.6:67,187,958 | 34 | 198 |
| | Hs06815519_cn | | Chr.6:67,379,660 | Probe excluded | |
| 7p12.2 | Hs05006345_cn | - | Chr.7:49,221,712 | 34 | 198 |
| | Hs04985951_cn | | Chr.7:49,293,487 | 34 | 24 |
| 9p23 | Hs03276415_cn | <i>PTPRD</i> | Chr.9:10,333,515 | 34 | 24 |
| | Hs03277406_cn | - | Chr.9:10,640,718 | 34 | 24 |
| | Hs06894124_cn | | Chr.9:10,996,224 | 34 | 24 |
| | Hs06871865_cn | | Chr.9:11,043,173 | 34 | 198 |
| 11p15.4 | Hs06324577_cn | <i>STIMI</i> | Chr.11:3,927,239 | 34 | 25 |
| | Hs06286180_cn | | Chr.11:3,940,908 | 34 | 198 |
| | Hs06291426_cn | | Chr.11:3,961,210 | 34 | 25 |
| 13q14.3 | Hs06378690_cn | - | Chr.13:54,280,419 | 34 | 198 |
| | Hs05292114_cn | | Chr.13:54,443,857 | 34 | 25 |
| 17p12 | Hs05513120_cn | <i>SHISA6</i> | Chr.17:11,453,820 | 34 | 198 |
| | Hs05498056_cn | <i>ZNF18</i> | Chr.17:11,887,582 | 34 | 24 |
| | Hs06420475_cn | <i>MYOCD</i> | Chr.17:12,594,654 | 34 | 24 |
| 17q22 | Hs03978967_cn | <i>C17orf67</i> | Chr.17:54,890,968 | 34 | 24 |
| | Hs03973468_cn | <i>DGKE</i> | Chr.17:54,942,334 | 34 | 198 |

Supplemental Table SII. Additional rare X chromosome CNVs identified but not followed up in patients with prune belly syndrome

| Locus | Genomic Coordinates | Size (bps) | Type | Study Subject ID | Gene(s)/Transcript(s) |
|--------------|----------------------------|-------------------|-------------|-------------------------|------------------------------|
| Xq13.2 | 71,784,938.. 71,957,183 | 172,245 | Dupl | 12 | <i>PHKA1</i> |
| Xq21.1 | 81,243,685.. 81,273,865 | 30,180 | Del | 12 | NA |

Abbreviations: Del: Deletion, Dupl: Duplication.
Coordinates (hg19) predicted using PennCNV.

Supplemental Table SIII. Phenotype of 14 prune belly syndrome cases with identified CNVs

| Study Subject ID | Phenotype |
|-------------------------|---|
| 1 | Hirschsprung, intestinal duplication, unspecified anomaly of gastrointestinal tract |
| 2 | Dextrocardia, imperforate anus |
| 3 | Isolated |
| 4 | Intestinal duplication, accessory kidney, absence of ureter |
| 5 | Ostium secundum, hip dysplasia |
| 6 | Isolated |
| 7 | Gastrointestinal tract malrotation |
| 8 | Gastroschisis |
| 9 | Isolated |
| 10 | Isolated |
| 11 | Isolated |
| 12 | Isolated |
| 13 | Isolated |
| 14 | Accessory kidney, pectus excavatum, omphalocele |