Figure S1. Overview of the study procedures. CNV, copy number variation; HLPA, high-throughput ligation-dependent probe amplification; WGS, whole-genome sequencing.
| Case number | CNV location | CNV length (kb) | Duplication/deletion | Genes involved | Heredity of CNVs | Clinical significance |
|-------------|--------------|----------------|----------------------|----------------|-----------------|----------------------|
| V130        | 9p24.3       | 334            | Duplication          | CBWD1, WASH1, FOXD4, DOCK8, FAM138C, C9orf66, DDX11L5 | V | US |    |
| V134        | 3q13.13      | 504            | Duplication          | -              | V | US | Suspected benign |
| V138        | 2p24.3       | 811            | Duplication          | -              | V | US | Suspected benign |
| V150        | 4q13.2       | 105            | Deletion             | UGT2B28        | V | US |    |
| V1001       | 11p15.1      | 320            | Duplication          | TPH1, KCNC1, SERGEF | V | US |    |
| V62         | 10q26.3      | 128            | Deletion             | LOC619207, CYP2E1, SYCE1 | V | US |    |
| V114        | 4p15.2       | 191            | Duplication          | C4orf52        | V | US |    |
| V133        | 15q11.2      | 130            | Deletion             | PWRN2          | V | US |    |
| Xq27.3      | 202          | 202            | Deletion             | -              | V | US | Suspected benign |
| V158        | 7q36.1       | 308            | Duplication          | ATP6V0E2-AS1, ZNF862, ATP6V0E2, DPP6 | V | US |    |
| V128        | 5q23.1       | 519            | Duplication          | -              | V | US | Suspected benign |
| V54         | 1p11.12      | 104            | Duplication          | LINC00273      | V | US |    |
| V68         | 22q11.22     | 272            | Duplication          | TOP3B, PPM1F   | V | US |    |
| V71         | 10q23.2      | 163            | Deletion             | LOC439994, LOC728190, FAM22A, FAM22D | V | US |    |
| V87         | 17q21.32     | 111            | Duplication          | HOXB-AS3, MIR196A1, MIR10A, HOXB2-9 | V | US |    |
| V105        | 16q24.3      | 110            | Deletion             | FANCA          | V | US | Pathogenic |
| V113        | 5p14.3       | 370            | Duplication          | INTS4L2, LOC441242, ZNF92 | V | US |    |
| V77         | 22q11.23q12.1| 263            | Deletion             | LRP5L, CRYBB2P1, IGLL3P | V | US |    |
| V79         | 1q31.1       | 213            | Deletion             | -              | V | US |    |
| 11q21q22.1  | 2.87 Mb      | 542            | Deletion             | -              | V | US | Suspected benign |
| 1q423.2     | 121          | Duplication     | SGPP1                | V | US |    |
| 15q13.3     | 519          | Duplication     | CHRNA7               | V | US |    |
Table SI. Continued.

| Case number | CNV location | CNV length (kb) | Duplication/deletion | Genes involved | Heredity of CNVs | Clinical significance |
|-------------|--------------|-----------------|----------------------|----------------|------------------|----------------------|
| V110        | Yp11.32-Yp11.2 | 7.91 Mb          | Deletion           | SHOX, PPP2R3B   | Paternal         | /                    | Pathogenic           |
| V122        | 4p16.3        | 3.8 Mb           | Deletion           | FGFR3, LETM1, WHSC1 | /                | /                    | Pathogenic           |
| V134        | 15q25.3       | 1.3 Mb           | Duplication        | AGBL1          | /                | /                    | VOUS                 |

CNV, copy number variation; VOUS, variants of uncertain significance.
| Case number | CNV position | Genes involved | Heredity of CNVs | Clinical significance |
|-------------|--------------|----------------|-----------------|-----------------------|
| V130        | 9p24.3: 10,001-344,129 (334 kb dup) | CBWD1, WASH1, FOXD4, DOCK8, FAM138C, C9orf66, DDX11L5 | ● | VOUS |
| V134        | 3q13.13: 110,249,785-110,753,998 (504 kb dup) | FAM86C2P | ● | Suspected benign |
|             | 11q13.2: 67,496,896-67,739,830 (242 kb del) | - | ● | VOUS |
|             | 15q11.2: 24,583,642-24,786,960 (203 kb del) | - | ● | Suspected benign |
| V138        | 2p24.3: 3,543,327-14,354,620 (811 kb dup) | - | ● | VOUS |
| V150        | 4q13.2: 70,138,883-70,244,272 (105 kb del) | UGT2B28 | ● | VOUS |
|             | 4q31.21: 144,929,449-145,037,725 (108 kb del) | GYPB, GYPA | ● | VOUS |
| V1001       | 11p15.1: 17,760,874-18,081,810 (320 kb dup) | TPH1, KCNC1, SERGEF | ● | VOUS |
|             | 15q11.2: 24,583,642-24,786,960 (203 kb del) | FAM86C2P | ● | VOUS |
|             | 12p13.33: 1,937,118-2,313,987 (376 kb dup) | - | ● | VOUS |
| V62         | 10q26.3: 135,247,534-135,376,327 (128 kb del) | LOC612907, CYP2E1, SYCE1 | ● | VOUS |
|             | 13q22.2: 75,964,191-76,285,851 (321 kb del) | TBC1D4, COMMD6, UCHL3, LMO7 | ● | VOUS |
| V114        | 4p15.2: 25,875,906-26,076,557 (191 kb dup) | C4orf52 | ● | VOUS |
| V133        | 15q11.2: 24,583,700-24,489,989 (130 kb del) | PWRN2 | ● | VOUS |
|             | Xq27.3: 143,134,313-143,337,405 (202 kb del) | - | ● | Suspected benign |
| V158        | 7q36.1: 149,556,685-149,864,823 (308 kb dup) | ATP6V0E2-AS1, ZNF862, ATP6V0E2, DPP6 | ● | VOUS |
|             | 7q36.2: 153,476,694-153,660,423 (183 kb dup) | LRP5L, CRYBBP2P1, IGLL3P | ● | VOUS |
|             | 2p11.23q12: 25,661,151-25,924,924 (263 kb dup) | - | ● | VOUS |
| V128        | q23.1: 120,345,246-120,864,255 (519 kb dup) | PRODH, DGCR6, USP18, GGT3P | ● | VOUS |
|             | 22q11.21: 18,644,144-18,914,454 (270 kb dup) | ZNF630, SPACA5B, SPACA5, SSX6 | ● | VOUS |
| V54         | 1p11.12: 48,850,055-48,954,577 (104 kb dup) | LINC00273 | ● | VOUS |
|             | 16p11.2: 33,946,471-34,183,059 (236 kb dup) | RAPI GAP2, OR3A1, OR1D2, OR1D5, OR1A1, OR1A2, OR1G1, OR1D4, OR3A2 | ● | VOUS |
| V68         | 22q11.22: 22,305,376-22,578,063 (272 kb dup) | TOP3B, PPM1F | ● | VOUS |
| V71         | 10q23.2: 88,966,588-89,130,520 (163 kb del) | LOC439994, LOC728190, FAM22A, FAM22D | ● | VOUS |
| V87         | 17q21.32: 46,609,588-46,720,700 (111 kb dup) | HOXB-AS3, MIR196A1, MIR10A, HOXB2-9 | ● | VOUS |
| V105        | 16q24.3 (89820001-89930000) (110 kb del) | FANCA | ● | Pathogenic |
| V113        | 5p14.3: 19,083,617-19,454,608 (370 kb del) | INTS4L2, LOC441242, ZNF92 | ● | Suspected benign |
|             | 7q11.21: 64,581,057-65,123,100 (542 kb del) | - | ● | VOUS |
| V77         | 22q11.23q12.1: 25,661,151-25,924,924 (263 kb del) | LRP5L, CRYBB2P1, IGLL3P | ● | VOUS |
| V79         | 1q31.1: 189,329,675-189,542,906 (213 kb del) | - | ● | Suspected benign |
|             | 11q21q22.195,882,357-98,752,576 (2.87 Mb del) | JRKL, MAML2, MIR1260B, CCDC82, JRKL-AS1 | ● | VOUS |
| Case number | CNV position            | Genes involved          | Heredity of CNVs | Clinical significance |
|-------------|-------------------------|-------------------------|------------------|-----------------------|
|             | 14q23.2: 64,177,898–64,299,096 (121 kb del) | SGPP1                   | /                | /                     | VOUS                  |
|             | 15q13.3: 31,994,610–32,514,175 (519 kb dup) | CHRNA7                  | /                | /                     | VOUS                  |
| V110        | Yp11.32-Yp11.2: 1-7,910,000 (7.91 Mb del)  | SHOX, PPP2R3B           | /                | /                     | Pathogenic            |
| V122        | 4p16.3: 68,345–3,916,139 (3.8 Mb del)      | FGFR3, LETM1, WHSC1     | /                | /                     | Pathogenic            |
| V134        | 15q25.3: 86,937,426–88,285,413 (1.3 Mb dup) | AGBL1                   | /                | /                     | VOUS                  |

CNV, copy number variation; VOUS, variants of uncertain significance; del, deletion; dup, duplication.