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This supplementary material has been provided by the authors to give readers additional information about their work.
eMaterial 1. Samples, genotyping methods and quality control (QC)
for the replication cohorts

Japanese sample

Sample description: The data from the Japanese sample are based on those reported by Ikeda et al, 2010 1: 1139 age- and sex-matched unrelated subjects of Japanese ethnicity (575 schizophrenic [SZ] patients and 564 control subjects). Control subjects were members of the general public who had no personal history of mental disorders. This was ascertained during face-to-face interviews where subjects were asked if they had suffered an episode of depression, mania, or psychotic experiences or if they had received treatment for any psychiatric disorder. Patients were entered into the study if they 1) met DSM-IV criteria for schizophrenia2; 2) were physically healthy and had normal routine laboratory tests; and 3) had no mood disorders, substance abuse, neurodevelopmental disorders, epilepsy, or known mental retardation. Consensus diagnoses were made by at least 2 experienced psychiatrists according to DSM-IV criteria2 on the basis of unstructured interviews with patients and families and review of medical records. Major medical conditions were obtained and body mass index was recorded based on available hospital records. After description of the study, written informed consent was obtained from each subject. This study was approved by the ethics committees of each participating university.

Genotyping and QC: All cases and controls were genotyped with Affymetrix 5.0 arrays. For the purpose of this study, we reanalyzed the raw data of that study. We used similar QC criteria as for the Bulgarian (BG) sample (except the z-score method). The total number of cases and controls in the Japanese sample was 490 and 516, respectively, after applying these filtering criteria.
Molecular Genetics of Schizophrenia (MGS)

Sample description: The MGS consists of cases and controls from European and African ancestry, described by Levinson et al., 2011. In total (after QC), there are 2671 cases and 2648 controls of European American ancestry and 1274 cases and 963 controls of African American ancestry. Details of the diagnostic methods have been described by Levinson et al., 2011; patients meeting DSM-IV criteria for SZ or schizoaffective disorder were included. Concomitant medical diagnoses were not available for this cohort.

Genotyping and QC: The specimens were assayed at the Broad Institute, Cambridge, Massachusetts, by using Affymetrix 6.0 genotyping arrays. Copy number variations (CNVs) were detected, or “called”, with the Birdseye module of the Birdsuite software package, version 2 (internal version 1.3), which uses a hidden Markov model algorithm. The data were normalized within plates of up to 92 DNA samples. HG18 human genome build locations are reported.

The CNV calls were merged if nearby pairs (or sequential pairs) of deleted or duplicated segments flanked a “normal” segment containing less than 20% of the probes in the merged CNV (primarily in segmental duplication regions). CNV calls were excluded if they overlapped (50%) with telomeres (100 000 base pairs [bp]) and centromeres, where CNV calls may be unreliable, or immunoglobulin gene regions where Epstein-Barr virus transformation causes structural changes. CNVs were also excluded if seen predominantly on 1 or 2 plates, suggesting artifact.

Samples were excluded based on 1) samples with total numbers of narrowly defined deletions or duplications exceeding the group mean by 3 SDs, 2) those with more than 2 chromosomes with outlier call numbers, 3) data for outlier chromosomes for subjects with 1 or 2 such chromosomes, and 4) samples (mostly lymphoblastic cell lines) with probe intensity variances exceeding the group mean by 4 SDs (predicting fewer CNV calls).
**International Schizophrenia Consortium (ISC)**

**Sample description**: The data from the ISC study are from 6 European populations and have been published before. The sample included **3045 cases and 3185 controls**, after excluding the BG cases that are probands in the trios from the current study.

**Genotyping and QC**: The samples were genotyped on Affymetrix 5.0 or 6.0 arrays at the Broad Institute. For the purpose of this study we examined the raw data and applied to it the same QC criteria as for the BG sample above, (except the $z$-score method).

**Vacic et al, 2011**

**Sample description**: The initial discovery data set was composed of 1761 unrelated subjects analyzed on the NimbleGen HD2 Array-CGH platform. The unfiltered sample consisted of 913 patients and 848 controls ascertained at 10 sites. Samples from the Trinity College Dublin cohort (n = 45) were removed for the purposes of this analysis, to avoid redundancy with the Wellcome Trust Case Control Consortium 2 (WTCCC2)/Irish sample (described below). The final discovery data set (after QC) consisted of **757 unrelated patients and 742 controls**.

**Genotyping and QC**: Microarray hybridizations using the NimbleGen HD2 platform were performed at the service laboratory of Roche NimbleGen according to the manufacturer’s specifications. All processing of microarray data was performed at Cold Spring Harbor Laboratory. Two segmentation algorithms were used to discover CNVs in the GC-corrected NimbleGen HD2 data: HMMSeg and Genome Alteration Detection Analysis (GADA; http://biron.usc.edu/~piquereg/GADA). CNVs detected by both algorithms were used for downstream data processing and analysis. CNVs detected by only 1 algorithm were excluded. In addition, CNVs of the same type (i.e., deletion or duplication) that were separated by ≤3
probes were merged into 1 contiguous segment. The proximal and distal boundaries of overlapping and adjacent CNVs were defined by the minimal chromosomal start position and the maximal chromosomal end position of the CNVs. CNVs with frequency >1% were removed. Rare CNV calls that passed the 1% frequency filter were further filtered based on the confidence score (CS). As the CS score of a CNV call we used the $P$ value derived from our outlier detection genotyping method, median $z$-score outlier detection (MeZOD), as previously published.\textsuperscript{10}

**WTCCC2/Irish sample**

**Sample description:** Controls in this sample were from 3 sources: 2663 persons from the 1958 British Birth Cohort and 2533 persons from the National Blood Service in Britain who were genotyped as part of the WTCCC2 study (http://www.wtecc.org.uk/ccc2/). The third control sample consisted of 979 Irish people obtained with written informed consent from the Irish GeneBank and represented blood donors from the Irish Blood Transfusion Service (IBTS) recruited in the Republic of Ireland. Blood donors were included in the study if they met ethnicity criteria (having 4 ethnically Irish grandparents). In Ireland and in the United Kingdom, blood donors are not screened formally for mental disorders, but people who report taking regular prescribed medication are excluded from donation. Donors are not financially renumerated. The 1958 Birth Cohort (also known as the National Child Development Study) includes all births in England, Wales, and Scotland, during 1 week in 1958. From an original sample of over 17 000 births, 9377 cohort members were visited at home for a biomedical examination at the age of 44-45 years (http://www.b58cgene.sgul.ac.uk/followup.php), and provided 7692 blood samples. The samples from these control cohorts are anonymized and we cannot obtain any further information about the medical or psychiatric histories of the carries with deletions.
The case samples were recruited through community mental services and inpatient units in the Republic of Ireland and Northern Ireland following similar research protocols and with local ethics approval. Diagnosis of a major psychotic disorder was made by the consensus lifetime best estimate method using DSM-IV criteria with all available information (interview, family or staff report, and chart review). The final number of subjects after QC filtering and relevant exclusions was 1269 patients (all from Ireland) and 6175 controls.

**Genotyping and Quality Control (QC):** All samples were genotyped using Affymetrix 6.0 arrays, and CNV calls were generated using Birdseye from Birdsuite, version 1.5.5.4 CNV calls were restricted to autosomal regions and standard QC included removal of CNVs of <100 kilobase (kb) and logarithm of odds score of <10. Common CNVs were excluded based on >50% overlap with a region that was copy number variable in at least 1% of samples. Individuals with more than 30 CNV calls or a total CNV sequence length of >10M bp were excluded, as well as any sample failing standard QC metrics based on single-nucleotide polymorphism (SNP) data.

**Swedish sample**

**Sample description:** All subjects were born in Sweden. Schizophrenia cases were identified via the Swedish Hospital Discharge Register containing all individuals hospitalized in Sweden since 1973, described in Bergen et al, 2012.11 Diagnoses were established by the attending physician and confirmed in a subset of subjects by medical record review. Cases, aged 18-65 years, must have had at least 2 hospitalizations with a SZ diagnosis, both parents born in Sweden, and signed written informed consent. Subjects were interviewed twice. A checklist for ancestry, somatic diseases (self-reported), and alcohol/drug use was completed. DNA was extracted from peripheral blood. Prevalence and recurrence risks for this definition
of schizophrenia are almost identical to that accepted by the field.\textsuperscript{12} A medical record review in the project showed 97\% concordance with DSM-IV criteria. Control subjects, also selected through registers, were group matched by age, sex, and county of residence. Controls had never received a discharge diagnosis of schizophrenia or schizoaffective disorder. All subjects were at least 18 years old and gave written informed consent to participate (however, we note that the control with a 16p11.2 deletion was recruited at the age of 45).

**Genotyping and QC:** Genotyping was performed using Genome-Wide Human SNP Array 6.0. Subjects and SNPs with genotype call rates <95\% were excluded. Population outliers assessed by multidimensional scaling were removed. One member of any pair of subjects demonstrating high relatedness (PI\textsubscript{HAT} >.20) was arbitrarily selected for removal. SNPs with marked departures from Hardy-Weinberg equilibrium ($P<1\times10^{-6}$) or very low minor allele frequencies (<1\%) were excluded. SNPs with nonrandom genotyping failure, inferred by the flanking haplotypic background using the PLINK\textsuperscript{13} mishap test, $P<1\times10^{-10}$, were removed from analysis. Plate-based associations of $P<1\times10^{-6}$ were taken as evidence of gross nonrandom plate failure based on a comparison of allele frequency of each plate to all others and were removed on a plate-by-plate basis. Intensity data from both SNP and CNV probes were used to identify CNVs based on a hidden Markov model.\textsuperscript{4} Only subjects who passed SNP quality control filters were considered for CNV analyses. CNVs with >1\% frequency, as well as common CNV regions indexed by HapMap, common rearrangements observed in blood, and any CNVs spanning large gaps (such as a centromere), were excluded. Analyses were restricted to autosomal CNVs greater than 100 kb. Seven subjects were removed for having either >10 Mb or >20 total CNVs. This resulted in 4429 CNVs (duplications:deletions ratio = 1.68) in 1506 schizophrenia cases and 2089 controls that remained for analysis (834 bipolar cases are not used in the current analysis).
German sample

A total of 1581 patients were included in the CNV analyses. These were recruited from consecutive admissions to psychiatric inpatient units and were all of German descent. Part of the sample (n = 487) was included in a previously published genome-wide association study (GWAS) and are described in detail as patient sample “GWAS Bonn-Mannheim” in Rietschel et al, 2011. A lifetime “best estimate” diagnosis of SZ or schizoaffective disorder according to DSM-IV criteria was assigned on the basis of the Structured Clinical Interview or the OPCRIT, medical records, and family history. Medical conditions relevant to psychiatric diagnosis were also recorded on the basis of these sources of information. After quality control, the sample consisted of 1386 patients with a male/female ratio of 1.14.

We used data from 1643 controls. Of these, 286 were recruited within the “Imaging Genetics” subproject of the German Integrated Genome Research Network “Systematic Investigation of the Molecular Causes of Major Mood Disorders and Schizophrenia.” None of these controls had a lifetime or family history of schizophrenia or any affective disorder. The remaining controls were drawn from 3 population-based epidemiological studies and are described in detail as control sample “GWAS Bonn-Mannheim” in Rietschel et al. After QC, the sample consisted of 1627 controls with a male/female ratio of 0.5.

Genotyping and QC: Venous blood samples were obtained from all participants. These were genotyped separately using the following Illumina BeadArrays: HumanHap550v3, Human610-Quadv1, and Human660W-Quad. Only those markers common to all 3 arrays were analyzed. SNPs with a call rate of < 97% were omitted and individuals were excluded from the dataset for the following reasons: 1) SNP call rate < 97%; 2) differences between X-
chromosomally inferred and phenotypic sex; 3) DNA sample doublets identified by identity-by-state estimates (defined as IBS = 2); 4) cryptic relatedness (IBS ≥ 1.6); and 5) population outliers according to multidimensional scaling with HapMap phase 2 data.

**CNV detection**: To identify potential CNVs, the SNP-chip data of each participant was analyzed using QuantiSNP (version 2.1, [http://www.well.ox.ac.uk/QuantiSNP](http://www.well.ox.ac.uk/QuantiSNP)) and PennCNV (version 2010May01, [http://www.openbioinformatics.org/penncnv/](http://www.openbioinformatics.org/penncnv/)). Participants were removed from the dataset if their SD from the log R ratio calculated over all SNPs exceeded 0.30.
eMaterial 2. 16p11.2 Probe Coverage

We examined the probe coverage of all arrays used in the current study, to ensure that the region of the 16p11.2 distal CNV is densely covered on each array type used in the studies, and therefore it is unlikely that any CNVs in this region could have been completely missed by some of the studies. The positions are shown in eFigure 3 and demonstrate a dense coverage of the region on all arrays.
eMaterial 3. Estimating the selection pressure against the distal 16p11.2 deletion

We were able to estimate the selection pressure (s) operating against deletions at distal 16p11.2 using the methods described in our previous publication.²⁴ Briefly, assuming a mutation-selection balance model, s should equal the deletion de novo rate [(de novo) / (inherited + de novo)] for this locus. We estimate the de novo rate, and therefore s, for deletions at distal 16p11.2 to be 0.25 (95% CI = 0.06-0.44), based on papers that have reported in a systematic way the rate of de novo and inherited CNVs in this region (eTable 3). This estimation is based on the observation of 5 de novos out of 20 systematically ascertained carriers from 5 available datasets, where inheritance had been examined (current study and refs 19-22). An s of 0.25 falls within the selection pressure range (0.12-0.88) of the other known schizophrenia-associated CNVs.²⁴
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eFigure 1. z-Score histogram for distal 16p11.2 (chromosome 16:28615687-28951365) in the Bulgarian trio data. Callout boxes indicate 2 transmitted deletion copy number variations, where the first 4 digits represent family ID and the last digit represents individual ID (1 = proband; 3 = mother).
**eFigure 2.** Principal component analysis plot of the Ashkenazi discovery cohort using 762,372 genome-wide autosomal single-nucleotide polymorphisms. Green dots represent case and blue dots represent control. As can be observed in the plot, there is minimal residual population structure in this cohort, and no evidence of batch effects.
**eFigure 3.** 16p11.2 region, including the known schizophrenia-associated locus (proximal) and the new locus implicated in the current study (distal). Shown are the positions of probes on the various arrays used in the current study.
eTable 1. Previous observations of distal 16p11.2 deletions. Further phenotypes tested for the 16p11.2 distal deletion. DD = developmental delay, ADHD = attention-deficit/hyperactivity disorder, SZ = schizophrenia, MCA = multiple congenital anomalies, MR = mental retardation. Note that the controls used in these studies are not independent from ours: Thus the Bochukova et al, 2010\textsuperscript{19} study used controls identical to those in the current study (Wellcome Trust Case Control Consortium 2 [WTCCC2] and Molecular Genetics of Schizophrenia [MGS]\textsuperscript{3}), the study by Bachmann-Gagescu et al, 2010\textsuperscript{21} used 3181 partially overlapping controls to those used in the current study (the International Schizophrenia Consortium ISC sample\textsuperscript{6}), and the study by Cooper et al, 2011 \textsuperscript{23} used some of the samples from WTCCC2. It is possible that all deletions in controls in these 3 studies are the same as those reported in the current study (from the WTCCC2 sample, where we report 2 deletions in the current study).

<table>
<thead>
<tr>
<th>Study</th>
<th>Phenotype</th>
<th>Platform</th>
<th>Case</th>
<th>Control</th>
<th>Comment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bochukova et al, 2010</td>
<td>Obesity/DD</td>
<td>Affy 6.0</td>
<td>300 (3)</td>
<td>&gt;7000 (2), (WTCCC2 + MGS)</td>
<td>143 cases had DD</td>
</tr>
<tr>
<td>Barge-Schaapveld et al, 2011</td>
<td>DD/unusual facial morphology/obesity.</td>
<td>aCGH Agilent.</td>
<td>Family 1 Two affected bothers carriers Family 2 Brother, Sister and Father affected carriers</td>
<td></td>
<td>Family 1–Father died early of paraproteinemia, very likely to have passed on deletion to both sons as mother does not carry the deletion.</td>
</tr>
<tr>
<td>Bachmann-Gagescu et al, 2010</td>
<td>Range of abnormal phenotypes, most commonly DD.</td>
<td>Multiple array platforms.</td>
<td>23 084 (31)</td>
<td>7700 (1). Controls come from 3 large published data sets.</td>
<td>Some patients with deletions have autism, ADHD, SZ, and Bipolar disorder</td>
</tr>
<tr>
<td>Bijlsma et al, 2009</td>
<td>MR/MCA</td>
<td>Various array platforms (distal deletion detected on Agilent 44k).</td>
<td>4284 (1)</td>
<td></td>
<td>Child with dismorpsisms and hypotonia. Father transmitting the deletion had learning difficulties.</td>
</tr>
<tr>
<td>Cooper et al, 2011</td>
<td>DD</td>
<td>Multiple array</td>
<td>15 767 (15)</td>
<td>8329 (1)</td>
<td></td>
</tr>
</tbody>
</table>
**eTable 2. Phenotypic data on carriers of 16p11.2 distal deletions.** Phenotypic data on carriers of 16p11.2 distal deletions, identified in the current study. Sz Undiff = Undifferentiated type of schizophrenia; Sz Par = paranoid type of schizophrenia; SzAff = schizoaffective disorder, Bip = bipolar type, dep = depressive type, sib = sibling, AAO = age at onset; AJ = Ashkenazi Jewish, BG = Bulgarian, EA = European American, MGS = Molecular Genetics of Schizophrenia.

<table>
<thead>
<tr>
<th>ID</th>
<th>Cohort</th>
<th>Sex</th>
<th>Ethnicity</th>
<th>Family History</th>
<th>Diagnosis</th>
<th>AAO, y</th>
<th>Course/clinical features</th>
</tr>
</thead>
<tbody>
<tr>
<td>82840</td>
<td>MGS</td>
<td>F</td>
<td>EA</td>
<td>grandparent SZ (not known if carrier)</td>
<td>Sz Undiff</td>
<td>24</td>
<td>Continuous</td>
</tr>
<tr>
<td>83897</td>
<td>MGS</td>
<td>M</td>
<td>EA</td>
<td>1 sib SZ (not known if carrier)</td>
<td>Sz Undiff</td>
<td>30</td>
<td>Episodic</td>
</tr>
<tr>
<td>72800</td>
<td>MGS</td>
<td>M</td>
<td>EA</td>
<td>1 sib SZ (not known if carrier)</td>
<td>Sz Undiff</td>
<td>15</td>
<td>Episodic (mild deterioration)</td>
</tr>
<tr>
<td>84406</td>
<td>MGS</td>
<td>F</td>
<td>EA</td>
<td>-</td>
<td>SzAff, Dep</td>
<td>16</td>
<td>Continuous; depressed ~50% of the time</td>
</tr>
<tr>
<td>97911</td>
<td>MGS</td>
<td>F</td>
<td>EA</td>
<td>-</td>
<td>Sz Undiff</td>
<td>15</td>
<td>Continuous</td>
</tr>
<tr>
<td>SZPBYK0003</td>
<td>AJ</td>
<td>F</td>
<td>AJ</td>
<td>-</td>
<td>Sz Par</td>
<td>30</td>
<td>continuous no medical problems noted</td>
</tr>
<tr>
<td>SZAABR0015</td>
<td>AJ</td>
<td>F</td>
<td>AJ</td>
<td>1 sib SZ (not known if carrier)</td>
<td>SzAff, Bip</td>
<td>25</td>
<td>Course data not available/ T2 diabetes, hypertension</td>
</tr>
<tr>
<td>P1336</td>
<td>Japan</td>
<td>M</td>
<td>Japanese</td>
<td>1 sib SZ (carrier)</td>
<td>Sz Par</td>
<td>26</td>
<td>2 episodes BMI=22.4</td>
</tr>
<tr>
<td>2220-1</td>
<td>BG trios</td>
<td>F</td>
<td>BG</td>
<td>Mother depressed (carrier)</td>
<td>Sz Catatonic</td>
<td>28</td>
<td>Acute onset, 1st episode, voices, paranoid, stupor, ideas of guilt</td>
</tr>
<tr>
<td>4290-1</td>
<td>BG trios</td>
<td>M</td>
<td>BG</td>
<td>Father anxiety (not carrier)</td>
<td>Sz Par</td>
<td>20</td>
<td>Continuous, voices, paranoid, self-neglect, “Overweight”</td>
</tr>
<tr>
<td>sbn-128-01</td>
<td>Germany</td>
<td>F</td>
<td>German</td>
<td>Father major depressive disorder (not known if carrier)</td>
<td>SzAff, Bip</td>
<td>23</td>
<td>No medical problems noted</td>
</tr>
<tr>
<td>Case-1</td>
<td>Sweden</td>
<td>F</td>
<td>Swedish</td>
<td>-</td>
<td>Sz Undiff</td>
<td>28</td>
<td>T2 diabetes, recurrent depression</td>
</tr>
<tr>
<td>Case-2</td>
<td>Sweden</td>
<td>F</td>
<td>Swedish</td>
<td>-</td>
<td>SzAff</td>
<td>24</td>
<td>Hypothyroidism, obesity</td>
</tr>
</tbody>
</table>

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eTable 3. Publications used to estimate the selection pressure(s) operating against deletions at distal 16p11.2, showing the numbers of reported de novo and inherited observations.

<table>
<thead>
<tr>
<th>Study</th>
<th>De novo/ (inherited + de novo)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bochukova et al\textsuperscript{19}</td>
<td>0/2</td>
</tr>
<tr>
<td>Bachmann-Gagescu et al\textsuperscript{21}</td>
<td>5/13</td>
</tr>
<tr>
<td>Current study</td>
<td>0/2</td>
</tr>
<tr>
<td>Barge-Schaapveld et al\textsuperscript{20}</td>
<td>0/2</td>
</tr>
<tr>
<td>Bijlsma et al\textsuperscript{22}</td>
<td>0/1</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>5/20</strong></td>
</tr>
</tbody>
</table>
eTable 4. CNVs in the Ashkenazi Jewish (AJ) and Bulgarian (BG) cohorts at loci previously implicated in schizophrenia. The list of loci is based on the reviews by Kirov, 2010 and Malhotra and Sebat, 2012 and represents only loci which we regard as having received strong statistical support, including replication evidence. CNV = copy number variant.

<table>
<thead>
<tr>
<th>CNV Locus</th>
<th>Position (Mb)</th>
<th>AJ cohort</th>
<th>BG cohort</th>
<th>Transmitted (de novo)</th>
<th>Not transmitted</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Case</td>
<td>Control</td>
<td></td>
<td></td>
</tr>
<tr>
<td>1q21.1 deletion</td>
<td>145.0-146.3</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>2p16.3 deletion (NRXN1)</td>
<td>50.0-51.1</td>
<td>0</td>
<td>1</td>
<td>4</td>
<td>1</td>
</tr>
<tr>
<td>3q29 deletion</td>
<td>197.2-198.8</td>
<td>1</td>
<td>0</td>
<td>1 de novo</td>
<td>0</td>
</tr>
<tr>
<td>7q11.23 duplication (WBS region)</td>
<td>72.4-73.8</td>
<td>0</td>
<td>0</td>
<td>1 de novo</td>
<td>0</td>
</tr>
<tr>
<td>7q36.3 duplication (VIPR2)</td>
<td>158.4-158.8</td>
<td>0</td>
<td>0</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>15q11.2 deletion</td>
<td>20.3-20.6</td>
<td>0</td>
<td>0</td>
<td>3</td>
<td>+ 2 de novo</td>
</tr>
<tr>
<td>15q11-13 duplication including PWS region</td>
<td>22.4-26.1</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>15q13.3 deletion</td>
<td>28.7-30.2</td>
<td>0</td>
<td>0</td>
<td>2 de novo</td>
<td>0</td>
</tr>
<tr>
<td>16p13.11 duplication</td>
<td>15.4-16.2</td>
<td>2</td>
<td>3</td>
<td>1</td>
<td>0</td>
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<tr>
<td>16p11.2 duplication</td>
<td>29.5-30.1</td>
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<td>0</td>
<td>1</td>
<td>+ 1 de novo</td>
</tr>
<tr>
<td>17p12/ deletion (HNPP)</td>
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<td>17q12 deletion</td>
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<td>0</td>
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<tr>
<td>22q11.21 deletion</td>
<td>17.4-18.7</td>
<td>2</td>
<td>1</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>
eReferences


13. Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MA, Bender D, Maller J, Sklar P, de Bakker PI, Daly MJ, Sham PC. PLINK: A tool set for whole-genome association and population-based linkage analyses. *Am J Hum Genet*. 2007; 81: 559-575.


