**Supplemental Data**

**Methods**

**DNA extraction and SNP microarray analysis**

Genomic DNA was extracted either automatically using the Chemagic™ Magnetic Separation Module I instrument  (PerkinElmer, Waltham, MA) or manually using Qiagen Puregene kits (Qiagen, Germany). The microarray analysis was performed using the Infinium Assay with the Illumina CytoSNP-850Kv1.2 BeadChip platform (Illumina, San Diego, CA). This chip contains approximately 846,500 genome-wide markers, an overall average probe spacing of 1.8 kb and an average effective resolution of 18 kb to determine copy number change. Copy number changes ≥10 SNP markers are reviewed for clinical significance. B-allele frequency and log2R ratio were analyzed with Illumina Genome Studio V2009.2 software. Results are described using the International System of Human Cytogenetic Nomenclature (ISCN: 2016). Linear positions of abnormalities are listed according to the Human Genome Build (GRCh37: Feb. 2009(hg19)). Information regarding genes located within chromosomal regions are obtained from the Database of Genomic Variants (http://projects.tcag.ca/variation/), the human genome browser at UCSC (http://genome.ucsc.edu/), and the NCBI RefSeq Project (http://www.ncbi.nlm.nih.gov/RefSeq).

**Fragile X analysis**

Isolated genomic DNA was assayed for CGG-repeat expansion of the *FMR1* locus by analysis of DNA fragments generated by the AmplideX FMR1 polymerase chain reaction (PCR) (Asuragen, Austin, TX) and separated by capillary electrophoresis ABI 3500XL Genetic Analyzer (Applied Biosystems, Foster city, CA).  Normal and mutation categories of FMR1 allele were determined according to the ACMG guidelines with normal repeat size as 5-44, gray zone as 45-54, premutation as 55-200, and full mutation >200.  For Southern blot analysis, DNA underwent restriction digestion followed by gel electrophoresis, after which it was transferred to a membrane (Thermo Fisher Scientific, Waltham, MA). Ultra-violet (UV) light was used to permanently fix the DNA onto the membrane which was probe hybridized, and then using chemiluminescence, it was visualized on an imager (Roche, Indianapolis, IN).

***PTEN* and *MECP2* PCR and sequencing**

The entire coding region and exon/intron boundaries of genes were analyzed by PCR and bidirectional sequencing for *PTEN* (NM\_000314.4) and *MECP2* (NM\_004992.2) according to manufactory instructions (Roche, Indianapolis, IN). The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed. Mutations in regulatory regions or other untranslated regions are not detected by this test. Large deletions involving entire single exon or multiple exons, large insertions and genetic recombinational events may not be identified using these methods. Detailed methodology is available upon request.

**Blood chromosomes**

Chromosomal analysis was performed according to standard procedures using GTG-banding. Peripheral blood lymphocytes were cultured in RPMI 1640 medium enriched with FBS, phytohemagglutinin and L- glutamine (Thermo Fisher Scientific, Waltham, MA). The cells were cultured for 72 hours at 37°C. Cells were harvested by adding colcemid for 2 hours. Cells were exposed to a hypotonic solution (KCl 0.075 mol/L) and fixed with methanol/acetic acid (3:1) (vol/vol). Metaphase chromosome spreads were prepared and G-banding with the use of trypsin-Giemsa at a resolution of 550 bands was established. A minimum of 20 metaphases were examined from each patient. Chromosome abnormalities were described according to the ISCN 2016.

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