Supplemental data

Supplemental Fig1. Boxplot analysis of the log2ratios from the 26 relevant BACs in control samples. The 7/26 BACs showing a high signal dispersion compatible false positive results are indicated by arrows; an asterisk indicates the loci that were not confirmed by additional methods (MLPA and/or oligo-aCGH)

Supplemental Table 1a. Description of the unique CNVs initially detected by the HSBA exclusively in several ASD patients, including the BAC ID, CNV location, size, nature, gene content, validation results and methods used for validation. We considered that two CNVs could be the same when they overlapped in more than 70% of their predicted genomic content.

Supplemental Table 1b. Description of all CNVs detected by the HSBA in ASD patients, including the BAC ID, CNV location, size, nature and gene content.

Supplemental Table 2. Candidate ASD genes located in the 13 CNV regions in 12 ASD patients with predicted functions and pathways. The interrelated pathways are shown in bold.

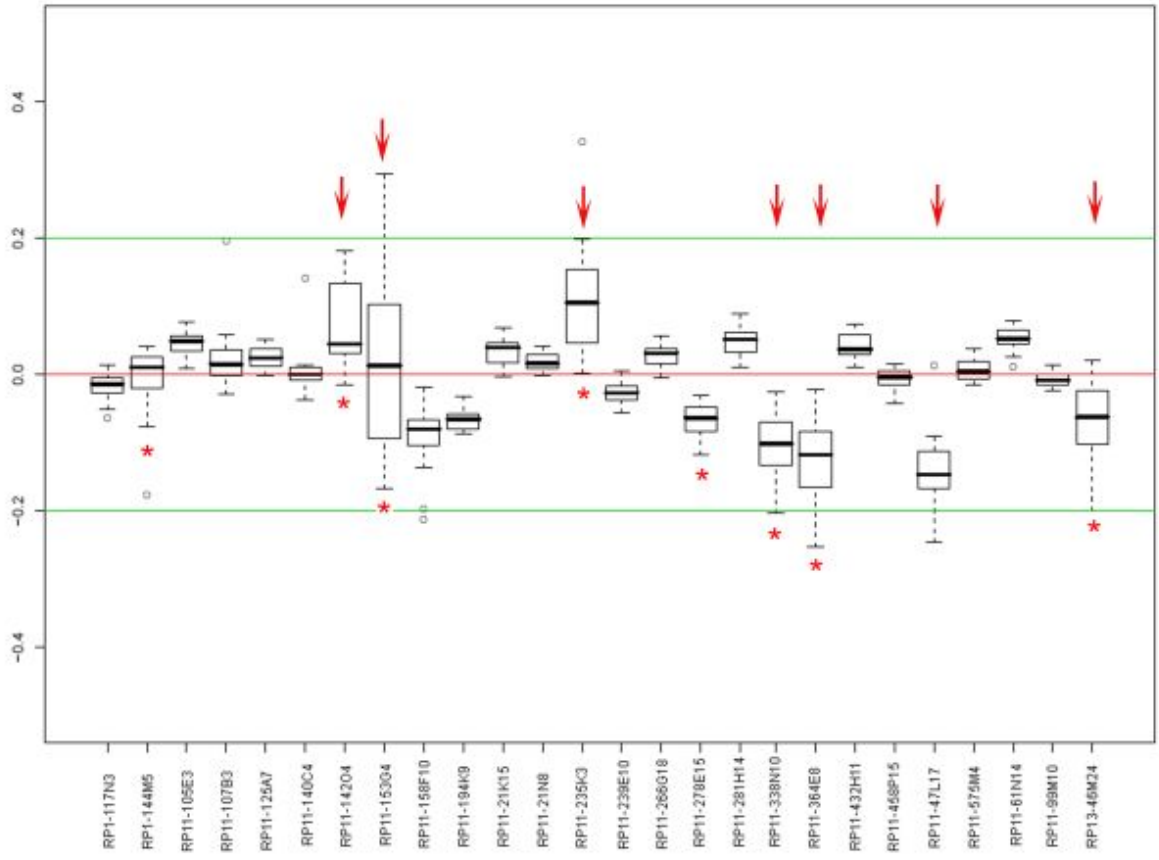
Supplemental Table 3. List of most represented pathways in the set of 24 genes located in the ASD-related CNVs in our cohort (analyzed with IPA, ConsensusPathDB and KEGG). The most relevant and interrelated pathways are marked in yellow.

Supplemental Table 4. List of most represented pathways among all genes located in ASD-related CNVs described by other investigators (analyzed with IPA, ConsensusPathDB and KEGG). The pathways in common with those found in the analysis of our sample are marked in yellow.

Supplemental Table 5. Full list of BAC clones included in the BAC array used (HSBA).

Supplemental Table 6. Sequence of the MLPA probes used for validation of the different rearrangements initially detected.

Supplemental Table 7. Differences in size estimation obtained with the HSBA BAC array and the oligo/SNP array for the ASD-specific confirmed CNVs.


0.4
0.2
0.0
-0.2
-0.4
RP1-117N3
RP1-144M5
RP11-105E3
RP11-107B3
RP11-125A7
RP11-140C4
RP11-142O4
RP11-153G4
RP11-158F10
RP11-194K9
RP11-21K15
RP11-21N8
RP11-235K3
RP11-238E10
RP11-266G18
RP11-278E15
RP11-281H14
RP11-338N10
RP11-364E8
RP11-432H11
RP11-458P15
RP11-47L17
RP11-575M4
RP11-61N14
RP11-99M10
RP13-465M24