

SUMMARY

The present inventors have performed genome wide association study on several large patient cohorts and have successfully identified a number of target genes harboring copy number variations associated with autism and ASD. Thus, in

5 accordance with the present disclosure, kits are provided for performance of a method for detecting a propensity for developing autism or autistic spectrum disorder is provided. An exemplary kit comprises means for obtaining a sample from a patient and testing the sample for the presence or absence of at least one deletion containing CNV in a target polynucleotide, wherein if the CNV is present, the patient has an
10 increased risk for developing autism and/or autistic spectrum disorder. In a preferred example, the deletion containing CNV is selected from the group of CNVs provided in Table II. In another example, the kit includes reagents for performing the step of detecting the presence of said CNV and further comprises specific reagents for performing a process selected from the group consisting of detection of specific
15 hybridization, measurement of allele size, restriction fragment length polymorphism analysis, allele-specific hybridization analysis, single base primer extension reaction, and sequencing of an amplified polynucleotide.

In one aspect, the present disclosure refers to a method for diagnosing a propensity for the development of autism or autistic spectrum disorder (ASD) in a
20 subject suffering from a 22q11.2 deletion syndrome, comprising:

detecting in a nucleic acid in a biological sample obtained from said subject, at least one CNV in a group of target genes, wherein the target genes are mGluR network genes, wherein, if a CNV deletion is present in at least one other mGluR network gene outside of the 22q 11.2 region, said subject has an increased risk for developing
25 ASD.

What is claimed is:

1. A method for diagnosing a propensity for the development of autism or autistic spectrum disorder (ASD) in a subject suffering from a 22q11.2 deletion syndrome, comprising:

detecting in a nucleic acid in a biological sample obtained from said subject, at least one CNV in a group of target genes, wherein the target genes are mGluR network genes, wherein, if a CNV deletion is present in at least one other mGluR network gene outside of the 22q 11.2 region, said subject has an increased risk for developing ASD.

2. The method of claim 1, wherein the mGluR network genes belong to the GRM family of genes.

3. The method of any one of the preceding claims, wherein the at least one CNV deletion is detected in a target gene selected from GRM1, GRM3, GRM5, GRM7 and GRM8.

4. The method of any of the preceding claims, wherein the ASD is syndromic ASD.

5. The method of any one of the preceding claims, further comprising a step of isolating said nucleic acids prior to detection.

6. The method of any one of the preceding claims, further comprising amplifying said group of target genes prior to detection.

7. The method of any one of the preceding claims, wherein said target genes are analyzed by performing a process selected from the group consisting of detection of specific hybridization, measurement of allele size, restriction fragment length polymorphism analysis, allele-specific hybridization analysis, single base primer extension reaction, and sequencing.

8. The method of any one of the preceding claims, wherein said target genes are

analyzed by performing a process selected from the group consisting of Southern and Northern blotting, direct sequencing and PCR amplification followed by hybridization of amplified products to a microarray comprising reference nucleic acid sequences.

9. The method of any one of the preceding claims, wherein the biological sample is a body fluid sample such as a blood sample, a urine sample, a serum sample or a gastric lavage sample, or a cell sample such as a sample of brain cells, of white cells or of mononuclear cells.