

SUPPLEMENT TABLE 1. Details on child and parent phenotyping and family classification

Child	<p>The exact screening procedures and measures for ASD phenotyping in children have been described in previous publications which can be consulted for greater detail (van Steijn et al. 2012). The parent and teacher Social Communication Questionnaire (SCQ) (Rutter et al. 2003) and the Child Social Behavior Questionnaire (CSBQ) (Hartman et al. 2006) were used to identify children with ASD symptoms. These questionnaires are validated instruments to measure ASD traits (Rutter et al. 2003; Charman et al. 2007). All children scoring above cut-off on any of the questionnaires underwent full diagnostic ASD assessment, including the Autism Diagnostic Interview-Revised (ADI-R) (Le Couteur et al. 2003). Control children were required to obtain non-clinical scores in order to be accepted in the study.</p>
Parent	<p>Case and control parents were screened for ASD using the Autism Spectrum Quotient (AQ) (Baron-Cohen et al. 2001) and the Adult Social Behavior Questionnaire (ASBQ) (Horwitz et al. 2005; Horwitz et al. submitted). The ASBQ is the adult version of the CSBQ and, although still under development, shows first promising results in terms of reliability and validity for the ASBQ (Horwitz et al. submitted). Parents scoring above cut-off (Hoekstra et al. 2008) on any of the questionnaires, were considered affected. Control parents were required to obtain non-clinical scores in order to be accepted in the study.</p>
Family classification	<p>Families were then stratified into SPX and MPX families. SPX families were required to have a single-affected proband, a minimum of one male sibling and all siblings and parents of the proband unaffected by ASD on the basis of non-clinical scores on the screenings questionnaires and/or administered diagnostic interviews. Families with siblings and/or parents who displayed (sub) threshold ASD symptoms, in addition to the proband, were categorized as multiplex (MPX). Families were excluded if a) only one unaffected parent from a presumed SPX family based on number of affected children participated in this study (to minimize the risk of erroneous categorization because of missing parental data) and b) if the affected proband had only female unaffected siblings (to account for higher sibling recurrence risk in male siblings than female siblings).</p>

Note. ASD = autism spectrum disorders; SPX = simplex; MPX = multiplex

References

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