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Identifying unique versus shared pre- and perinatal risk factors for ASD and ADHD using a simplex-multiplex stratification

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

Child	The exact screening procedures and measures for ASD and ADHD 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), the Child Social Behavior Questionnaire (CSBQ) (Hartman, Luteijn, Serra, & Minderaa, 2006), and Conners Rating Scales Revised (CPRS; CTRS) (Conners, 1996) were used to identify children with ASD and/or ADHD symptoms. These questionnaires are validated instruments to measure ASD and ADHD traits (Charman et al., 2007; Conners, Sitarenios, Parker, & Epstein, 1998; Rutter et al., 2003). All children scoring above cut-off on any of the questionnaires underwent full diagnostic ASD and ADHD assessment, including the Autism Diagnostic Interview-Revised (ADI-R) (Le Couteur, Lord, & Rutter, 2003) and Parental Account of Childhood Symptoms ADHD subversion (PACS) (Taylor, Sandberg, Thorley, & Giles, 1991). Control children were required to obtain non-clinical scores in order to be accepted in the study.
Parent	In the ASD cohort, case and control parents were screened for ASD using the Autism Spectrum Quotient (AQ) (Baron-Cohen, Wheelwright, Skinner, Martin, & Clubley, 2001) and the Adult Social Behavior Questionnaire (ASBQ) (Horwitz, Systema, Ketelaars, & Wiersma, 2005). 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). In the ADHD cohort, case and control parents were screened for ADHD using the self-report questionnaire for ADHD (Kooij et al., 2005), the self and spouse Conners Adult Rating Scales Self Report (CAARS:S-L) (Conners, Erhardt, & Sparrow,

	1998, 1999), and the Schedule for Affective Disorders and Schizophrenia for School-Age Children - Present and Lifetime Version (K-SADS-PL; administered in follow-up study [NeuroIMAGE]) (Kaufman et al., 1997). Parents scoring above cut-off (Hoekstra, Bartels, Cath, & Boomsma, 2008; Kooij et al., 2005) on any of the ASD/ADHD questionnaires, or on the semi-structured ADHD interview, were considered 'affected'**.** Control parents were required to obtain non-clinical scores in order to be accepted in the study.
Family classification	Families were then stratified into SPX and MPX families, see Oerlemans, Hartman, de Bruijn, Van Steijn et al., 2015, Figure 1. 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 or ADHD 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 and/or ADHD 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).

Note. ASD = autism spectrum disorders; ADHD = attention-deficit/hyperactivity disorder; SPX = simplex; MPX = multiplex

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