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The underlying symptom structure of autism spectrum disorders: A factor analytic approach using the developmental, dimensional and diagnostic interview

Wouter De la Marche [a,b], Ilse Noens [a,c,d], Bart Boets [a,e], Sofie Kuppens [f], Jean Steyaert [a,e,g,*]

[a] Leuven Autism Research (LAuRes), KU Leuven (University of Leuven), Belgium
[b] Youth Department, Public Psychiatric Care Centre Geel, Belgium
[c] Parenting and Special Education Research Unit, KU Leuven (University of Leuven), Belgium
[d] Psychiatric and Neurodevelopmental Genetics Unit, Massachusetts General Hospital, Boston, MA, USA
[e] Child and Adolescent Psychiatry Department, KU Leuven (University of Leuven), Belgium
[f] HIVA – Research Institute for Work and Society, KU Leuven (University of Leuven), Belgium
[g] Clinical Genetics, Maastricht University Hospital, Netherlands

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ABSTRACT

Several studies have focused on the underlying symptom structure of Autism Spectrum Disorders (ASD), but results have been equivocal. We performed a confirmatory factor analysis on data of the Developmental, Dimensional and Diagnostic Interview of 275 participants with ASD between 3 and 23 years of age, aimed at strengthening the empirical evidence of previously published factor structure solutions using the same instrument. As none of these hypothesised models fitted our data, an exploratory factor analysis was undertaken. Results pointed towards a five factor model. A 'Restricted and Repetitive Behaviour and Interest' factor could be separated from 'Shaking and Nodding', 'Emotional Reciprocity' and two other factors that both represented deficits in social interaction and communication. Although not completely confirming, our results are generally in favour of the present DSM-5 criteria. By showing that the items did not fully segregate according to theoretically postulated subdomains, we offer a possible explanation for the heterogeneity in proposed factor structures for ASD.

Abbreviations: ASD, autism spectrum disorder(s); So, social deficits (as criterion of pervasive developmental disorders in DSM-IV-TR); Com, deficits in communication (as criterion of pervasive developmental disorders in DSM-IV-TR); SoCom, deficits in social communication (as criterion of ASD in DSM-5, as underlying construct for ASD in different papers); RRBI, restricted and repetitive pattern of behaviour and interests; CFA, confirmatory factor analysis; EFA, exploratory factor analysis; PCA, principal component analysis; FSIQ, full scale intelligence quotient.

* Corresponding author at: Child and Adolescent Psychiatry Department, UPC-KU Leuven campus Gasthuisberg, Herestraat 49, B-3000 Leuven, Belgium. Tel.: +32 16 343821; fax: +32 16 343830.
E-mail addresses: wouter.delamarche@opzgeel.be (W. De la Marche), jean.steyaert@uz.kuleuven.be (J. Steyaert).

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1. Introduction

With every new version of the Diagnostic and Statistical Manual (DSM), criteria have changed for the condition currently referred to as autism spectrum disorder (ASD). Changes pertain to several aspects, such as the number of key domains of impairment, the number and content of the individual criteria, the number of criteria needed for diagnosis, as well as the supposed interrelation between individual criteria. These changes are evident in the transition from the former DSM-IV-TR (American Psychiatric Association, 2000) to the new DSM-5 (American Psychiatric Association, 2013): most criteria of the formerly distinct domains on social (So) and communication (Com) impairments have been adapted and reorganised into one domain, namely social communication and social interaction (SoCom); criteria on imitation and imaginative play on their own have disappeared, as well as language delay; idiosyncratic and repetitive use of language have moved to the domain of restricted, repetitive patterns of behaviour, interests, or activities (RRBI); and a criterion on atypical sensory processing was added to the latter domain. Though all these changes are based on research and clinical experience, they have stirred a storm of discussion about the sensitivity, specificity and inclusion/exclusion range of the new DSM-5 diagnosis (as reviewed by Halfon & Kuo, 2013).

In preparation for and parallel with the development of DSM-5, clinicians and researchers have been looking for the most appropriate way to capture and describe the underlying constructs of ASD. From a clinical point of view, it is interesting to know which symptoms generally co-occur and how they are related to other domains of functioning (such as cognitive abilities). This may be particularly relevant to guide therapeutic interventions and follow-up. From a scientific point of view, delineating more stable symptom clusters in specific subsamples can be of great importance to determine specific neurocognitive, biological and genetic precursors of (specific subgroups of) ASD. This is what drives the search for informative phenotypes (Szatmari et al., 2007).

Conducting factor analysis or related statistical methods is a widely used strategy to determine underlying constructs of ASD. While confirmatory factor analysis (CFA) is used to assess the extent to which a hypothesised structure – resulting from theory, previous empirical findings, or both – provides a good fit to the data, exploratory factor analysis (EFA) is used to explore the underlying structure of the observed variables at hand (Pett, Lackey, & Sullivan, 2003). These approaches have been applied to data from various standardised diagnostic instruments for ASD: questionnaires, parent interviews and observation schedules. The most commonly used questionnaire in this regard is the Social Responsiveness Scale (SRS; Constantino & Gruber, 2005). Factor analytic studies have also been conducted on two diagnostic interviews and one observation schedule. The semi-structured Autism Diagnostic Interview-Revised (ADI-R; Lord, Rutter, & LeCouteur, 1994), comprising a diagnostic algorithm in which DSM-IV-TR criteria are operationalised, is the most widely used. The Developmental Dimensional and Diagnostic Interview (3di; Skuse et al., 2004) is a semi-structured interview, also based on the DSM-IV-TR criteria. It is computerised, and while related items of behaviour are lumped into symptom scores in the ADI-R, the 3di addresses these items separately. The Autism Diagnostic Observation Schedule (ADOS; Lord et al., 2000) is the most widely used observation instrument in factor analytic research.

To give a brief overview of previous factor analytic studies on the ASD phenotype, we performed two PubMed searches up to March 2014 using the terms "autism AND factor structure" and "autism AND factor analysis", resulting in 138 and 477, partially overlapping, papers. These papers were then screened on title and abstract. Studies were included if they (a) described a factor analysis or related technique; (b) of the autism phenotype; (c) in an ASD sample; (d) using instruments that were described in at least two studies. We excluded (a) studies that merely focused on instrument construction and thus not purely on the underlying constructs of ASD; (b) studies that solely targeted a general population or broad child psychiatric sample; (c) studies using instruments that were not focusing on core ASD behaviour (e.g., neurocognitive and biological characteristics, co-morbid disorders and symptoms); and (d) studies based on an instrument that was only used once.

Table 1 offers an overview of the 20 papers that met these criteria, ordered by research instrument, and including characteristics of the conducted analyses and main results. Researchers have used different statistical methods to gain insight into the underlying structure of the ASD phenotype. EFA and CFA are the most commonly used techniques (see supra for a description). An orthogonal rotation such as varimax assumes that the factors are uncorrelated, while in an oblique rotation the factors are allowed to correlate, which may better approach clinical reality but may also hamper interpretation. Principal axis factoring is the most widely used method in factor analysis; it adopts iterative algorithms for factor extraction. Principal component analysis (PCA) is another technique, which aims to reduce the observed variables into a smaller number of principal components that maximally accounts for the total variance in the observed variables. Although factor analysis and PCA are both data reduction techniques, they are mathematically different because PCA includes the total variance (unique and common), while factor analysis only uses the common variance. Conceptually, with factor analysis the researcher also assumes that an underlying causal model exists, whereas PCA is simply a linear transformation of the initial data vectors (Pruzek, 2005). While factor analysis assumes latent dimensions or factors, latent class analysis assumes the existence of discrete latent categories of classes. Moreover, factor mixture is a hybrid model that combines latent class analysis and common factor analysis allowing one to model latent factors across classes (Lubke & Muthèn, 2005).

Factor analytic studies on questionnaires have yielded very different results depending on the particular instrument used. In their first factor analytic studies using the SRS, Constantino et al. (2004) have advocated that ASD could be seen as a unitary construct. Other authors, however, have suggested a two-factor model based on a combination of items of the SRS and the Social Communication Questionnaire (SCQ; Rutter, Bailey, & Lord, 2003), with the first factor combining both social