Autism spectrum disorder: Fractionable or coherent?

David M Williams and Dermot M Bowler

Autism 2014 18: 2
DOI: 10.1177/1362361313513523

Editorial

autism

Autism spectrum disorder: Fractionable or coherent?

Autism
2014, Vol 18(1) 2–5

DOI: 10.1177/1362361313513523
aut.sagepub.com
SAGE

The initial impetus for this special issue was a landmark article by Happé and Ronald (2008; see also Happé et al., 2006), in which it was claimed that, contrary to widespread belief, autism spectrum disorder (ASD) is not a coherent syndrome in the classical sense. Instead, it was hypothesised that the behavioural deficits in social interaction, communication and behavioural flexibility (restricted and repetitive behaviour and interests) that were diagnostic of ASD in *Diagnostic and Statistical Manual of Mental Disorders–Fourth Edition, Text Revision* (*DSM-IV-TR*; American Psychiatric Association (APA), 2000) are separable, with each feature having independent psychological/cognitive, neurobiological and genetic bases. As such, these features can occur separately, but only when they co-occur can the ASD be diagnosed. Of course, this view is somewhat at odds with the new diagnostic framework laid down in *Diagnostic and Statistical Manual of Mental Disorders–Fifth Edition* (*DSM-5*; APA, 2013), which characterises ASD in terms of a dyad (rather than triad) of behavioural deficits in social-communication and behavioural flexibility. The collapsing of social interaction deficits and communication deficits is not in accord with the view that these behavioural features are separable, although it does not rule out the possibility that they have distinct underlying causes.

One reason why the idea of a fractionable triad was potentially revelatory was because it provided a new and possibly more tractable way to approach the understanding of the causes, as well as the management, of ASD. Despite almost half a century of research into all aspects of ASD, there are still no reliable diagnostic biomarkers for ASD, and nor do we know enough about how to remediate the core features of the disorder either through medication (see Blankenship et al., 2011), as Rutter notes in his commentary, or through behavioural interventions (e.g. Green et al., 2010). If Happé and Ronald's (2008) hypothesis is correct, then research (and intervention efforts) should be directed towards understanding each component feature of ASD in isolation, rather than ASD as a whole.

What, then, do the articles in the special issue contribute to this debate? Since ASD is currently a behaviourally defined condition, a good place to start this debate is to establish the relative coherence of the different diagnostic features of the disorder. The articles by Mandy et al. and Frazier et al. broadly support the structure proposed by *DSM-5* by finding that a two-factor solution fitted the data that they obtained using the Developmental, Dimensional and Diagnostic Interview (3di; Skuse et al., 2004), and the Social Responsiveness Scale-2 (SRS; Constantino and Gruber, 2012), respectively. Particularly reassuring was that Mandy et al. found this two-factor solution in both UK and Finnish sample of individuals with ASD, suggesting cross-country/cross-cultural reliability in the structure of core ASD features. Both these sets of findings can be considered congruent with some notion of fractionation among the features of ASD, in that neither Frazier et al.'s study or Mandy et al.'s study found that a one-factor model adequately fitted their data. However, it is striking that in both studies, the two factors were associated significantly. In Frazier et al.'s study, the associations between SRS scores in social-communication domain and SRS scores in the repetitive and restricted behaviour domain were remarkably high, ranging from .87 to .95 in their ASD sample, child general population sample and adult general population sample. Although the SRS is somewhat weighted more towards measuring social-communication features than repetitive and stereotyped behaviour (in that the latter aspect of the dyad is assessed by only a small proportion of the 65 items that comprise the SRS), it is striking that the association between the two aspects of the dyad is so strong in each of these samples. The sheer size of the samples also leaves little room to question the reliability of the findings, which is a clear strength of the data reported by Frazier et al. In Mandy et al.'s study, the association between the two factors was smaller in magnitude, but notably consistent across their UK sample (r = .43) and Finnish sample (r = .44). These observations raise the question of what should count as coherent when considering ASD features? Both studies suggest strongly that the two core ASD features are potentially *separable*, but not that they are *unconnected*.

A further important finding by Mandy et al. is that subclinical manifestations of ASD-like features among individuals with the 'broad autism phenotype' do not possess the same structure among a Finnish sample as they do among a UK sample. The importance of the broad autism phenotype to the debate about whether ASD features cohere is that some individuals appear to have significant ASD-like traits, but only in one domain. This is taken as prima facie evidence that the features of ASD are fractionable. However, Mandy et al.'s article may suggest that

observations of people with ASD may not generalise to individuals with the broad autism phenotype, which may lead to caution when considering the applicability of the broad autism phenotype to the debate about whether ASD is fractionable. As Rutter points out in his commentary, a definitive answer to these questions must lie outside a consideration of the co-occurrence of behaviours across individuals, however, important this approach might be.

For the claim that autism is fractionable, it is not enough simply to document patterns of behaviour. Two further things are needed: first, a conceptual analysis of what would count as coherent or fractionable in this context, and second, an analysis of data relating to factors thought to *underlie* the behavioural features. The article by Brunsdon and Happé addresses these requirements by exploring the extent to which the cognitive causes of ASD are fractionable. The claim that ASD is fractionable would be strengthened if it could be demonstrated that the cognitive underpinnings of the disorder (a) are independent of each other and (b) contribute to independent behavioural features of the disorder. If it could be shown that each behavioural feature of ASD had an entirely distinct underlying cause, then this would contradict the notion that the two features are fundamentally coherent (although there would still be scope for the features to share causes at other levels of explanation, or share a developmental relation). Based on their comprehensive review of the literature, Brunsdon and Happé conclude that there is partial evidence to support each of these points/considerations. In particular, they argue that the evidence points to deficits in theory of mind as a contributory cause of social-communication features of ASD (but not repetitive and restricted behaviour) and executive dysfunction (and, to a lesser extent, weak central coherence) as a contributory cause of repetitive and restricted behaviour (but not social-communication features). Potentially, this is highly important, because, among other things, it suggests that cognitive-based interventions will need to target individual cognitive deficits *in combination* if they are to successfully ameliorate all aspects of ASD. However, Brunsdon and Happé also highlight some contradictory evidence, which reflects the inconsistent pattern of findings in the existing literature. There are two difficulties, in particular, when trying to interpret these findings.

The first potential difficulty is that (as argued by Hobson, in his article) only one of the cognitive accounts of ASD that Brunsdon and Happé consider – the Theory of Mind (ToM) account – is well specified. 'Executive functioning' is an umbrella term for several (separable, but related) functions that are involved in the control of action (e.g. Miyake et al., 2000). It is not clear which aspect/component of executive functioning is (proposed to be) a unique and specific contributory cause of ASD features, nor whether a specific profile of executive dysfunction explains features of the disorder. Indeed, some investigators question whether such broad notions as 'executive dysfunction' can adequately explain specific forms of developmental psychopathology, such as ASD (see, for example, Morton, 2004; but see Russell, 1996). Similarly, although the construct of 'weak central coherence' is highly useful at a descriptive level, it is still not clear what mechanism might underpin a pattern of behaviour that confers coherence at levels as disparate as perceptual and conceptual processing. Thus, in our view, the notion of weak central coherence as presently construed can take us only a limited way forward in our understanding of the basis of ASD.

The second, and perhaps more fundamental, difficulty comes when trying to distinguish cognitive task performance from the cognitive *mechanisms* that underpin performance. As Brunsdon and Happé rightly point out, cognitive tasks are rarely if ever 'process pure'. This means that it is difficult to be certain whether associations (or lack thereof) between cognitive task performance and behavioural features result from (a) an underlying association (or lack thereof) between the cognitive process of interest (e.g. ToM) and behaviour, or from (b) an underlying association (or lack thereof) between extraneous, non-specific task processes (e.g. verbal intelligence) and behaviour. The same is true when considering associations (or lack thereof) between performance on two or more tasks that assess different cognitive constructs (e.g. ToM tasks and executive functioning tasks). This difficulty is compounded in the case of ASD, given that good performance on cognitive tasks can be achieved via compensatory mechanisms despite limited underlying cognitive competence. One hallmark of compensation, unlike cognitive competence, is that it will be inconsistent across different situations. Thus, it is striking that Brunsdon and Happé found some degree of (even if not total) consistency in associations between behavioural features of ASD, and performance on a variety of tasks that purport to measure ToM, executive functioning and central coherence, respectively. This makes it more likely (but not definitively the case) that specific cognitive deficits in ASD *are* related to (possibly specific) behavioural features in ASD, which highlight the importance of Brunsdon and Happé's review and of why, more generally, we need to study cognition in ASD if we are to fully understand the disorder (Frith, 2012; Morton and Frith, 1995).

The question remains, however, to what extent do these findings rule out the possibility that ASD represents a coherent syndrome? In his article, Hobson provides a valuable conceptual analysis of what would count as evidence for fractionation or coherence, as well as evidence for his conviction that ASD can be considered a coherent disorder. As part of his conceptual analysis, he questions whether the measures that have been used to identify the behavioural and genetic structure of ASD are sufficiently sensitive (and reliable) to capture the qualities of ASD features – and specifically 'ASD-like' features among individuals from the general population. Hobson makes several important points

about the data that Happé and Ronald (2008) use to support their fractionation account, but his general point is that apparent fractionation (or, in fact, coherence) of ASD-like features may be an artificial consequence of insensitive measures of ASD features, rather than genuine fractionation. In his commentary, Rutter makes a similar point about the sensitivity of measures to the broad autism phenotype. As such, this point also has relevance for interpretation of the finding of Mandy et al., reported in this issue, that there are cross-cultural differences in the coherence/structure of ASD/ASD-like features among individuals with the broad autism phenotype. It is possible that these findings reflect merely an insensitivity of the measure they used to assess features (the 3di), rather than any genuine differences in coherence of features across cultures. However, some reassurance that Mandy et al.'s findings are not merely an artefact of insensitive feature measurement is provided by the fact that the 3di is a clinician-administered measure. This means that the trained interviewer has the opportunity to prompt for relevant information and to interpret the answers given by interviewees in relation to the qualities of ASD features. Thus, the 3di may be sensitive to the qualities of ASD/ASD-like features to a degree that questionnaire measures are not (although this does not guarantee that the measure is sensitive to the qualities of the broad autism phenotype, as Rutter notes). In general, though, we believe the challenges posed by Hobson (and by Rutter) are not to be taken lightly, even if there turns out to be a tractable solution to them.

As part of his argument that ASD *is* a coherent syndrome, Hobson suggests that a cognitive/psychological deficit in the ability to 'identify' with the bodily expressed attitudes of other people represents a final common pathway to the syndrome of ASD as behaviourally defined. He points out that congenitally blind children merit diagnoses of ASD at rates far above chance/the population estimate. Presumably, ASD among these individuals does not have the same genetic (or perhaps neurobiological) cause(s) as it does among sighted children who receive a diagnosis. Rather, Hobson argues, ASD among congenitally blind and sighted children has the same cognitive/psychological cause, namely, a reduced ability to identify with others. In blind children, this diminution results from 'non-central' damage to the visual system, which disrupts *input* to the otherwise-intact cognitive/psychological 'identification' mechanism, whereas in sighted children, it results from 'central' damage to the mechanism itself. Whether or not one accepts Hobson's claim regarding the shared cognitive causes of ASD in sighted and blind children, what is clear is that ASD features themselves cohere, rather than fractionate, among blind children with a diagnosis. If one accepts the reasonably uncontroversial claim that ASD among blind children does not have the same aetiology as it does in sighted children, then this example strikes a blow to the notion that ASD features in sighted children are merely a chance co-occurrence.

As readers might expect, the four articles and commentary in this special issue are not going to settle the question of whether ASD, as we currently understand it, is a disorder that is best conceptualised as a coherent whole or as a co-occurring set of sub-features. We are encouraged that, as the contributions to this special issue highlight, there is considerable debate about the answer to this question. What has been established is that there is a refreshing reliability about the identification and classification of the behavioural features of ASD. This, at least, suggests that individuals on the spectrum can be reliably differentiated from those who are not. Moreover, it points to a certain coherence at least on the behavioural level. Quite where we go when we travel beyond the behavioural level is more problematic and depends in part on an individual scientist's or clinician's preoccupations and training. In his commentary, Rutter quite rightly notes the importance of considering issues of genetics, of neurological impairment such as epilepsy, and of treatment studies. We ourselves, as developmental and experimental psychologists, will inevitably be drawn to the conceptualisation of underlying cognitive and more general psychological mechanisms, as well as their operationalisation. The articles in this special issue show that there are still important questions to be answered regarding the level of analysis we should adopt (genetic, neural, psychological or behavioural) when speaking about ASD, and particularly whether the categories that are meaningful at one level are easily translated to another. A syndrome that is coherent at the biological level might be radically fractionable at the behavioural level, or *vice versa*. Rutter's and Hobson's mention of diabetes is informative here. Originally, 'diabetes' meant 'excessive discharge of urine', which together with characteristic excessive thirst, forms a reasonably coherent syndrome. However, we now know that there are several types of diabetes. Type 1 diabetes results from insulin deficiency consequent on pancreatic dysfunction, whereas type 2 diabetes results from insulin resistance rather than deficiency. Both produce symptoms of excessive urination and increased thirst (and thus make a coherent syndrome), but the underlying biochemistry and pancreatic pathology differ. A disorder we now know to be unrelated to these two – *diabetes insipidus* – shares the same symptoms of increased urination and thirst, but results from disordered kidney-related metabolism. Thus, three conditions that are coherent at the 'behavioural' level (urination and thirst) require radically different biological explanations and interventions.

Finally, there is the issue of development. It is ironic that although ASD is universally characterised as developmental in nature, development has to a large extent been overlooked in the contributions to this special issue. However, as Rutter rightly argues, we must consider development seriously, because ASD features may become more or less inter-connected over time. Thus, our conclusion about the relative fractionation or coherence of the disorder may

differ according to when in development we explore the issue. Rutter cites Pellicano (2013) as suggesting that initially fractionated traits might become more coherent with development as a consequence of shared contextual effects in development. The alternative is also possible, namely, that initially coherent, inseparable traits fractionate over time. This could occur as a result of compensation in one domain (e.g. learning not to engage in stereotypies), but not another domain. This would give the superficial impression of incoherence of features, despite coherence in their original manifestation. The central point is that we should never forget that the clinical picture we see among individuals with a diagnosis of ASD represents a particular point in an atypical developmental trajectory, in which both the clinical features and any putative underlying factors may be in a process of change. The challenge to understand how this process of change operates will no doubt add an additional layer of complexity to the picture, but we have little doubt that this layer will be necessary to resolve the debate about fractionation/coherence.

All of these considerations matter, because all of them impact in one way or another how we envisage our scientific and clinical encounters with people on the autism spectrum. This will differ depending on whether we see all of ASD as a set of manifestations of a single or small set of gene or brain dysfunctions, or whether we see different observable patterns of behaviour as emergent results of the interplay of different psychological processes. Whether and how we choose to intervene will, of course, depend also on a wider consideration of the individuals themselves, their carers, and their families. We hope that this special issue serves to sharpen our focus on these issues and will contribute to the lively ongoing debate about the coherence or otherwise of ASD.

References

American Psychiatric Association (APA) (2000) *Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR)*. 4th ed. Text Revision. Washington, DC: American Psychiatric Association.

American Psychiatric Association (APA) (2013) *Diagnostic and Statistical Manual (DSM-5)*. 5th ed. Washington, DC: American Psychiatric Association.

Blankenship K, Erickson CA, Stigler KA, Posey DJ, et al. (2011) Psychopharmacological treatment of autism. In: Amaral D, Dawson G and Geschwind D (eds) *Autism Spectrum Disorders*. New York: Oxford University Press, pp. 1196–1214.

Constantino JN and Gruber CP (2012) *The Social Responsiveness Scale Manual (SRS-2)*. 2nd ed. Los Angeles, CA: Western Psychological Services.

Frith U (2012) Why we need cognitive explanations of autism. *Quarterly Journal of Experimental Psychology* 65: 2073–2092.

Green J, Charman T, McChonachie H, et al. (2010) Parent-mediated communication-focused treatment in children with autism (PACT): a randomised controlled trial. *Lancet* 19(3795): 2152–2160.

Happé F and Ronald A (2008) The 'fractionable autism triad': a review of evidence from behavioural, genetic, cognitive and neural research. *Neuropsychological Review* 18: 287–304.

Happé F, Ronald A and Plomin R (2006) Time to give up on a single explanation for autism. *Nature Neuroscience* 9: 1218–1220.

Miyake A, Friedman NP, Emerson MJ, et al. (2000) The unity and diversity of executive functions and their contributions to complex 'frontal lobe' tasks: a latent variable analysis. *Cognitive Psychology* 41(1): 49–100.

Morton J (2004) *Understanding Developmental Disorders: A Causal Modelling Approach*. Malden, MA: Blackwell.

Morton J and Frith U (1995) Causal modelling: a structural approach to developmental psychopathology. In: Cicchetti D and Cohen DJ (eds) *Developmental Psychopathology, Theory and Methods*, vol.1. New York: Wiley, pp. 357–390.

Pellicano E (2013) Testing the predictive power of cognitive atypicalities in autistic children: evidence from a 3-year follow-up study. *Autism Research* 6: 258–267.

Russell J (1996) *Agency: Its Role in Mental Development*. Hove: Erlbaum.

Skuse D, Warrington R, Bishop D, et al. (2004) The developmental, dimensional and diagnostic interview (3di): a novel computerized assessment for autism spectrum disorders. *Journal of the American Academy of Child and Adolescent Psychiatry* 43(5): 548–558.

David M Williams and Dermot M Bowler
Editors