

Title:

Autism: one or many spectrums?

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Autism: one or many spectrums?

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This is the author manuscript accepted for publication and has undergone full peer review but has not been through the copyediting, typesetting, pagination and proofreading process, which may lead to differences between this version and the Version of Record. Please cite this article as doi: [10.1111/jpc.14176](https://doi.org/10.1111/jpc.14176)

Abstract

Our conceptualisation of Autism Spectrum Disorder has changed over time, with recent classifications reflecting a heterogeneous clinical presentation now regularly encountered in routine general paediatric practice. As the prevalence of autism and associated demands for services have increased, so has research into understanding cause and trials aimed at providing best care and intervention. The heterogeneity of autism has meant that no single aetiology can account for all differences in presentation, however, and not all children benefit from broad-based interventions. Now is the time to rethink how best to understand individual differences, in order to focus research efforts and take steps towards more sophisticated strategies that go beyond the behaviours we look for when making an autism diagnosis. We suggest adopting a dimensional approach to autism assessment, with consideration of eight spectrums of abilities, ways of thinking and behaviour. This eight spectrum approach will assist clinicians to consider each individual's strengths and needs, and personalise interventions and support accordingly. Profiling individual skills across these dimensions may also provide researchers with greater capacity to link causal pathways with specific phenotypes, which is needed to develop precision medicine for autism.

The world has changed its view of autism since first described by Kanner in his seminal paper in 1943.¹ In Kanner's original account of 'autistic disturbances', he expressed the need for 'detailed consideration of (its) fascinating peculiarities'.¹ Over the years, however, there has been a shift from providing in-depth descriptions of presentation to defining individuals as having autism or not.

Once considered a rare disorder,² recent reports estimate the prevalence worldwide to be between 1 and 4%.³⁻⁶ Associated with this increase in diagnosis is the changing definition of autism, from a discrete mental health disorder with severe impact on functioning,⁷ to a neurodevelopmental disorder, with a broad spectrum of strengths and difficulties.

Several important changes have brought us to where we are today. Difficulty categorising individuals according to diagnostic classifications for pervasive developmental disorders in the Diagnostic and Statistical Manual of Mental Disorders Fourth Edition (DSM-IV)⁷ and 10th Revision of the International Classification of Diseases (ICD-10)⁸ led to subgroups being subsumed under the umbrella term Autism Spectrum Disorder in the most recent versions of both classification systems.^{9,10} Using DSM-5, autism diagnosis is behaviourally based, characterised by difficulties in social communication and reciprocity, and repetitive, stereotyped behaviours.⁹ The variation in behaviour that is consistent with diagnostic classification means that autism is not one thing. For example, a person with autism could be verbose and struggle to maintain social roles and employment given his unusual interaction style, or have little intelligible speech and require full time support to access learning in the classroom.

Over the decades there has also been a shift in social culture, with increasing community awareness and changing socio-political landscape for disability advocacy.^{3,6} This in turn has had an impact on the way autism is perceived.

What has changed for children and their families?

It can be argued that increased awareness and associated diagnostic trends have helped children to access support, as well as offering an explanation for the presence of ‘eccentric’ or challenging behaviours in some individuals. Increasing awareness of salient social communication differences as opposed to deficits has also provided opportunities for advocacy groups who do not want to be “fixed” to campaign for social change and improved public understanding. This movement is in line with the social model of disability, which recognises that society’s response to difference either creates or ameliorates disability.

On the other hand, increasing awareness has put pressure on clinicians and assessment teams to see many more children with only minor changes in resource allocation following Medicare item number changes in 2008, as part of the Helping Children with Autism Package (HCWA).¹¹ This has reduced opportunities for more detailed investigation into profiles at an individual level. Additionally, service funding dependent on diagnosis, rather than needs, has driven practice toward answering diagnostic, rather than needs-based, questions in Australia.

How do we move towards precision medicine for children with autism?

Understanding cause

A significant body of research has focused on uncovering a unifying cognitive or neurobiological cause for autism, but has failed to do so to date. Numerous cognitive theories have been proposed, including theory of mind deficits,¹² weak central coherence,¹³ executive function difficulties,¹⁴ and differences in empathising and sympathising as part of the extreme male brain theory.¹⁵

Researchers have also attempted to identify biological causes with wide-reaching theories linked to genes, inflammation, metabolic differences and neuroimaging abnormalities.¹⁶⁻²¹ Genetic explorations, historically focused on twins²²⁻²⁴ and children with known genetic conditions with an increased risk of autism,^{25,26} have recently identified common variant genes linked to other conditions and potential epigenetic differences, which fit with gene-environment interaction theories currently being explored.²⁷

Given the inability for any one cognitive theory to account for all differences in presentation²⁸ and the identification of many genes linked to autism²⁹ it is now proposed that, rather than a single cause, there are multiple pathways for multiple conditions ('autisms') which share phenotypic similarities consistent with a diagnosis of autism, but reflect the heterogeneity of the disorder.^{30,31} This conceptualisation is, however, not adequately explained by our current diagnostic classification systems.

Our understanding of autism or 'autisms', may be advanced through the Research Domain Criteria (RDoC) Framework, an approach developed by the National Institute of Mental Health (NIMH), which classifies mental disorders according to dimensions of observable behaviour and neurobiological measures which cut across the traditional diagnostic categories.^{32,33} The goal of the RDoC is to build a research literature that reflects

advances in genetics, neuroscience and behavioural science to provide a foundation for precision diagnosis. The RDoC conceptually aligns with a bio-psychosocial model of autism, taking into account multiple factors in understanding cause and identifying need. The RDoC model, designed as a matrix, acknowledges both the importance of monitoring change over time, with the inclusion of developmental trajectories, and the influence of environmental effects.

In brief, there are five domains within the RDoC Framework (negative valence, positive valence, cognitive, social processes, and arousal and regulatory) related to emotion, cognition, motivation, and social behaviour.³² Within each domain there are constructs and sub-constructs; for example, perception and understanding of self and others are two of four constructs within social processes relevant to autism. A goal of RDoC is to focus on relevant systems and their underpinnings, to document unfolding trajectories and interactions with events across the lifespan.

Best care and effective interventions

Happé and colleagues have drawn attention to how the heterogeneity of autism challenges our capacity to understand and evaluate trajectories and outcomes from intervention, noting that it is unlikely that all individuals with autism will benefit from the same types of approaches.³⁰ Indeed, despite best research efforts, controversy remains about the most effective interventions for children with autism.³⁴⁻³⁷ In their review of early intervention research, Stahmer *et al.*³⁸ reported that it is not uncommon for up to 50% of children to show substantial gains following intervention, while the remaining 50% show limited to modest gains in skill development. Questions therefore remain regarding what the

right intervention looks like for different children at different stages of development.

Researchers focused on outcomes following early intervention for children with autism are investigating ways to predict response to intervention by looking at both (a) pre-intervention child characteristics,³⁹⁻⁴¹ and (b) program elements,⁴² with the view to individualising care based on a child's initial presenting profile and supports needs.

The International Classification of Functioning (ICF), published by the World Health Organization (WHO) in 2001, is an established framework aiding conceptualisation of health conditions such as autism that can assist with best clinical care and outcomes relevant to assessing response to intervention.⁴³ The ICF framework describes influences on the expression of a condition, including contextual and personal factors, and highlights the importance of the interplay with function and participation. In 2014, a series of studies and consensus meetings were initiated to develop ICF "Core Sets" for autism, to specify the important categories and factors that are influential across the spectrum.⁴⁴ At a recent consensus meeting, 111 categories were identified for inclusion in the Comprehensive Core Set, arguably reiterating the heterogeneity of autism.⁴⁵ Many categories fell within environmental, activity and participation domains, with relatively few relating to body structure and function; most related to mental function.⁴⁵ This is in keeping with the shift away from the medical model of conceptualising autism, towards the more recent social models of disability. Brief core sets were also defined for different age groups, highlighting variation in the presentation of autism across the lifespan.

How will services provide needed support?

In 2008, the Australian Government established the HCWA Package for children diagnosed with autism aged under 7 years.⁴⁶ The HCWA package provided families the opportunity to access assessments and interventions (including resources), were equal in value irrespective of severity of autism symptoms, place of residence, or service availability, and were not adjusted for family income. Currently, the HCWA funding model is being absorbed into the National Disability Insurance Scheme (NDIS). The NDIS differs from the HCWA package in that the scheme is designed to provide access to services based on individualised plans that have been developed based on an individual's function-based difficulties and diagnosis.⁴⁷ The NDIS is still in initial phases of implementation, with scarce data available as yet in relation to true practice change.⁴⁷ Some early evidence suggests that whilst there is an increased emphasis on the functional needs of the individual under the NDIS, a person's diagnosis and associated severity level at the time of assessment remain critical components of resource planning and allocation.⁴⁸ A model that requires clarification of important domains of development alongside function may best assist the NDIS to provide the right service for the right child at the right time.

Do current diagnostic approaches provide enough for informing intervention choices?

Despite clear guidance from the WHO⁹ and the National Institute for Health and Care Excellence (NICE),⁴⁹ the autism diagnostic process varies widely in Australia^{50, 51} and overseas⁵² and is not always linked to broader neurodevelopmental and functional profiling assessing both strengths and difficulties. Within Australia, this assessment process is currently under review.⁵³ A number of standardised instruments have been developed to

inform a diagnosis of autism, with the Autism Diagnostic Interview-Revised (ADI-R)⁵⁴ and the Autism Diagnostic Observation Schedule–Second Edition (ADOS-2)⁵⁵ most widely used. These assessments have been designed to elicit and assess behaviours characteristic of autism, but provide little in the way of informing prognosis or intervention planning. There are also concerns regarding the sole reliance on these standardised measures for informing a diagnosis, particularly for preschool age children and children with lower cognitive abilities.^{56,57} Professional judgement and clinical experience therefore play a substantial role in the accurate identification of behavioural criteria associated with autism and forming of a clinical diagnosis.^{58,59} Implicit in this diagnostic process is our reliance on a clinician's subjective understanding of what behaviours are considered “acceptable” compared with those which are clinically significant.

An eight spectrum approach

How do we provide children and families with the information they need to access personalised care?

Like others we propose a dimensional, rather than diagnostic, framework for assessment of each child's strengths and difficulties.^{33,60} Crucial additional considerations specific to children include their vulnerability to their environment and associated ability to learn, and the likelihood that their behaviours will change over time. These factors need to be incorporated into assessment so that identified abilities and behaviours can be considered

within the context of an individual's environment, and care can be modified to align with changes over time.

We recommend clinicians and researchers consider each individual according to eight dimensions of ability, thinking and behaviour (see Figure 1). These dimensions can be used to highlight strengths and difficulties in a way that has potential to support individuals and families in decision making about best interventions and future planning needs and aligns with the NDIS initiative of providing choice and control for its participants. We have adapted a model proposed by the USA Government Accountability Office,⁶¹ by adding level of intelligence, to allow assessment of behaviours related to cognitive ability as recommended in DSM-5, and dimensions of attention/focus and symptoms of anxiety/mood difficulties, to reflect characteristics common among individuals with autism.⁶²⁻⁶⁴ Within each dimension, the full spectrum of ability or behaviour is considered. To illustrate, for concentration and focus the range is from limited ability to concentrate (which could be consistent with an attention deficit) to high levels of concentration to the exclusion of other things. Once strengths and difficulties within the eight dimensions are identified, their potential impact on behaviour, functional difficulties and participation can be assessed. For example, a need for routine could affect social interactions, levels of anxiety and sensory-seeking behaviours in specific contexts, such as when a relief teacher is in a classroom. In turn, this information can be used to identify priority areas and suitable targets for intervention.

Within this eight spectrum approach, an individual can be assessed against established population norms and expectations on some dimensions. Assessment against norms, when available, can assist with diagnostic formulation, with sufficient deficits in the right

dimensions indicating diagnostic justification. To illustrate, a comprehensive language assessment could aid in differential diagnosis when identifying a language, rather than social communication disorder. We propose that a dimensional model will add value to existing practice of norm-referenced testing by also highlighting areas of absolute or relative strength and identifying possible co-morbidities.

A dimensional approach also aids conceptualisation of how an individual's profile can fluctuate over time, change across settings and be considered an asset or a hindrance across contexts.⁶¹ An individual's ability to maintain focus on small detail may be well suited to computer activities, for example, but less useful when needing to get ready for school in a timely fashion. In this way, dimensional information can be used in clinic for discussing strategies for specific settings in which more or less focus is needed.

We acknowledge that within each proposed dimension there is complexity and that the dimensions interact to influence each other. Intelligence is not one thing, e.g. some individuals present with verbal comprehension difficulties and others with low processing speed. Similarly, some individuals are resistant to some changes, like their room being rearranged, but enjoy large variety in their diet. Some individuals show sensory sensitivity to particular textures, but tolerate variation in other senses, such as loud noises. Language abilities are likely to influence performance on intelligence testing, and both will influence social interaction. The tools used for assessing each dimension may assist in identifying differences within other dimensions that have implications for function, support and intervention, including approaches to education. An understanding of within-dimension

variation, across age groups and contexts, may also trigger the need for additional assessments that may change intervention and education strategies.

How can clinical information contribute to research advances?

In clinical care we are not able to assess all the elements suggested by RDoC. However, improved documentation of dimensional strengths and difficulties that enables more personalised care could also be used by researchers to develop a greater understanding of how profiles vary in type and degree from person to person, with the possibility of identifying patterns among subgroups. As this approach develops it is likely that identifying cause will be linked to specific strengths and difficulties, rather than broad diagnostic categories.

Conclusion

The increasing heterogeneity of autism poses challenges both in research and practice. We need to better understand the underpinnings of autism, and develop tailored and comprehensive strategies for each child and family. We suggest that before diagnosing children as part of one broad spectrum, we need to understand their ability, ways of thinking and behaviour within eight individual spectrums, or dimensions. In so doing we can assist children and families to access the most appropriate interventions and supports for their specific strengths and needs, and understand how interventions and supports may change between settings and over time. Moreover, if we adopt an eight spectrum approach in

research we are likely to advance our understanding of causes. By doing this we will be one step closer to precision medicine for children with autism.

Acknowledgements

We thank the William Collie Trust, University of Melbourne and the Lorenzo and Pamela Galli Charitable Trust for their support to the authors A Ure, V Rose and K Williams.

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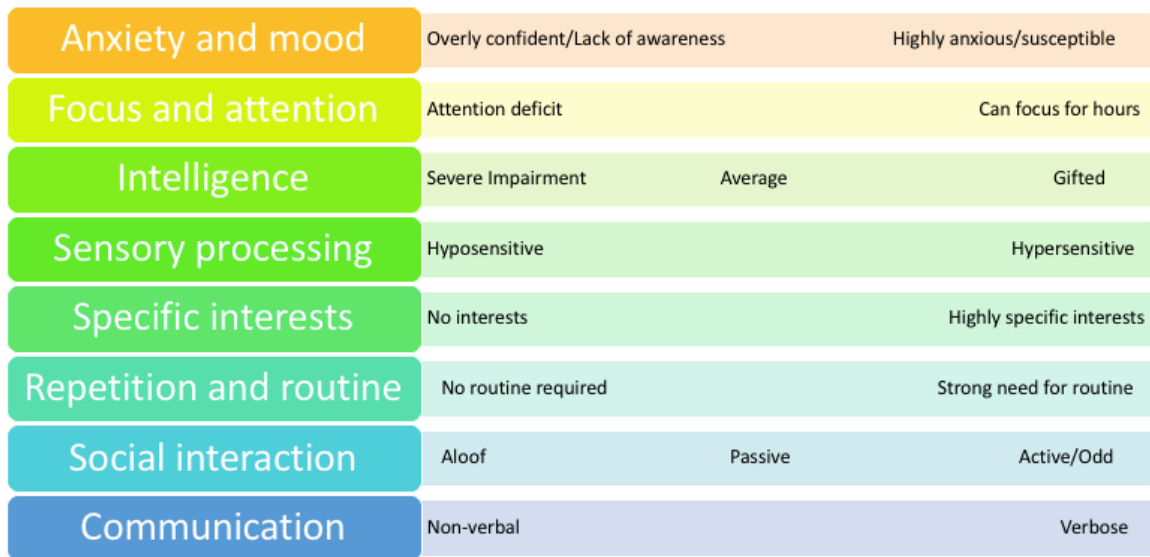


Figure 1. Eight spectrum approach to autism spectrum disorder



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2018-10-01

Citation:

Ure, A., Rose, V., Bernie, C. & Williams, K. (2018). Autism: One or many spectrums?. JOURNAL OF PAEDIATRICS AND CHILD HEALTH, 54 (10), pp.1068-1072. <https://doi.org/10.1111/jpc.14176>.

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