



Changes in the concept of autism

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Abstract

The conceptualisation of Autism has greatly evolved over the past several decades. In this review article, we focus on several areas where our understanding of Autism has changed: (1) from a 'narrow' definition to a 'wide' diagnostic criteria; (2) from a rare to a relatively common condition, although probably still under-recognised in women and older people; (3) a condition diagnosed predominately in males to now being identified in people of all genders; (4) from something affecting children, to a lifelong condition; (5) from something discrete and distinct, to a dimensional view; (6) from one thing to many 'Autisms', and a compound or fractionable condition; (7) from a focus on 'pure' Autism, to recognition that complexity and comorbidity is the norm; and finally, (8) from conceptualising Autism purely as a developmental disorder, to recognising a neurodiversity perspective, operationalised in participatory research models. We also explore opportunities for how research can become more generalisable, including in a global context, and make suggestions for areas currently neglected in Autism research.

Introduction

Autism spectrum disorder (henceforth simply 'Autism') is a neurodevelopmental condition characterised by difficulties with social relationships and social communication, rigid and repetitive behaviours and interests, and atypical sensory processing ¹. By current diagnostic criteria, autism is a relatively common condition, affecting approximately 1-2% of the population ². In terms of aetiology, autism has been found to be genetically influenced, with heritability being estimated to be over 80% ^{3,4}. Features of Autism often begin to emerge within the first two years of life (although in many cases, they may not be obvious or identified until later life ⁵) and persist across the lifespan ⁶. Further to the core features of Autism, autistic people¹¹ often experience a range of cognitive, emotional, and behavioural atypicalities. As such, complexity and co-occurrence is often the rule rather than the exception, with the most commonly co-occurring difficulties being intellectual disability, attention-deficit/hyperactivity disorder (ADHD), anxiety, speech and language problems, difficulties with emotional and behavioural regulation, and executive dysfunction ⁷⁻¹⁰. However, Autism is also characterised by a range of strengths; whilst Autism has no consistent associated pattern of cognitive peaks and troughs, a non-specific unevenness of abilities across cognitive domains is a hallmark of the condition ¹¹, with many autistic people excelling at detail-focused tasks ¹².

However, Autism has not always been viewed in this way. First described by Grunya Sukhareva in the 1920s, and later named and conceptualised by Hans Asperger and Leo Kanner in the 1940s, Autism (as a standalone condition unrelated to schizophrenia or psychosis) was not included in diagnostic manuals as until the 1980s. Since then, and particularly in the past decade, the diagnostic criteria and clinical description of Autism have evolved substantially. This article will reflect on the evolving conceptualisation of Autism, how we arrived at the condition we recognise today, and the opportunities for the future.

¹¹In this article, we have opted to use identity-first language, i.e., 'autistic people', rather than person-first language, i.e., 'people with Autism'. Many English-speaking people in the autistic community prefer identity-first language and consider it to be less stigmatising. We acknowledge that there may be different preferences for those who speak [French/Spanish], but have opted to use this approach as it is widely accepted in English, which this article was originally written in.

Autism has changed from 'narrow' to 'wide'

Initially, Autism was narrowly defined, but over time, the understanding has broadened to include a spectrum of diverse manifestations and experiences. For example, Autism was first labelled as 'Infantile Autism' in the American Psychiatric Association's 3rd edition of the Diagnostic and Statistical Manual (DSM-III¹³). 'Infantile Autism' had six narrow criteria, including a "pervasive lack of responsiveness to other people" and "gross deficits in language development", and could only be diagnosed in childhood. The narrow focus of these diagnostic criteria led to Autism being identified predominately in those with comorbid intellectual disabilities (present in ~75% of those diagnoses circa DSM-III), and most young people with this diagnosis would have been educated in specialist schools.

However, in the current 5th edition of the Diagnostic and Statistical Manual (DSM-5-TR)¹ criteria for 'Autism Spectrum Disorder', the diagnostic criterion for social functioning difficulties are framed more broadly as "persistent deficits in social communication and social interaction across multiple contexts...". Additionally, Autism can be diagnosed with or without language and intellectual impairments, and in people of all ages. Autism is now predominately being identified in those without comorbid intellectual disabilities (present in ~25% of those diagnosed circa DSM-5-TR), and many of the people identified are being supported in mainstream schools or living and working independently.

A consequence of this widening is that most Autism research now focuses on those without intellectual impairments, with a systematic review suggesting that over 90% of autistic people involved in research have an IQ in the normal range, and 80% of studies have selection biases against those with intellectual disabilities¹⁴. Therefore, the generalisability of research is reduced, and public perceptions of Autism may be skewed towards those without impairment. This may also impact how policy decisions are being made if the evidence base excludes those who are likely to need the most support.

Autism has changed from rare to common

An improved understanding of Autism has resulted in what was once considered a rare diagnosis, now being a common one. This improved understanding is marked by a substantial increase in identification rates. The first population estimates of 'autistic conditions' in the 1960s suggested that only 4 in 10,000 children (0.04%) would meet diagnostic criteria¹⁵. Lorna Wing then suggested in the 1980s that Autism may form a spectrum, with 22 in 10,000 children (0.22%) meeting criteria¹⁶. Identification rates have increased in the years since, with current estimates suggesting that between 1-3% of the population are autistic². The US Centre for Disease Control (CDC) reported in 2008 that the prevalence rate in 8-year-olds was 1/88, which was a 74% increase from their report in 2004. This increase was also noted in other countries, such as a fivefold increase in the UK between 2004 and 2010¹⁷. However, in recent years, this prevalence rate has stabilised, with little variation across different geographic regions¹⁸.

When considering why this rise in Autism diagnoses has occurred, it is important to remember several influencing factors. First, awareness of Autism has increased both clinically and in the general population, resulting in more people being identified as autistic. Second, the diagnostic criteria for Autism have widened, resulting in those once overlooked only now being identified (including those first diagnosed in adulthood). Finally, diagnostic substitution has occurred, where people once diagnosed with other conditions (e.g., personality disorders, etc.) are now having their diagnoses replaced with an Autism diagnosis^{19,20}. These points taken together suggest that improved identification rates may be driving this increase in Autism diagnoses, rather than a change in the actual prevalence of the condition²¹.

Autism has changed from mostly male to all genders

The shift in perspective now recognises that Autism affects individuals of all genders, challenging the historical emphasis on males. In the 1980s, the sex ratio of Autism was estimated at ~5-10 males to 1 female. Furthermore, when a woman/girl was identified as autistic during this time period, they often had a comorbid intellectual disability. However, recent estimates from epidemiological studies find that the sex ratio is less unbalanced, being 3 males to 1 female, and with similar rates of comorbid intellectual disabilities²².

The historic emphasis on autistic men and boys has also had an impact on how Autism is viewed, including

theories of the condition. For example, the ‘extreme male brain’ account was a leading theory for several years ²³, however, it is not as favourably viewed today ²⁴. Furthermore, the concept of ‘camouflaging’ has become a popular notion, where some autistic people (particularly women and girls) may hide their autistic characteristics by mimicking the behaviours of their neurotypical peers ^{25,26}. In addition, many people still have an implicit male bias or stereotype of autism; seeing a boy struggling socially they may think ‘Is he autistic?’, but seeing a girl struggling socially they may think ‘Is she shy? Does she have social anxiety?’. Research has indicated that women and girls are less likely to receive an Autism diagnosis when presenting similar symptom severity to men and boys ²⁷. Women and girls also often receive their Autism diagnoses much later in life compared to men and boys, meaning that many women and girls are undiagnosed and unsupported ²⁸. Furthermore, sex biases in ‘diagnostic overshadowing’ are seen; for example, a girl may receive a diagnosis of anorexia, but the clinician fails to look further and see she is also autistic ²⁹. As such, it is important that clinicians have an awareness of how these biases can manifest in their clinical practice.

Moving beyond sex to gender, another point to note is that many autistic people are gender diverse (i.e., their gender does not match their sex assigned at birth) ³⁰. Approximately 10% of autistic people experience gender dysphoria, compared to 3% in the general population ³¹. The rates of non-traditional gender identities (e.g., non-binary, gender fluid) are also higher amongst autistic people than their neurotypical peers. Greater awareness is needed of how autistic traits and characteristics may manifest in people of different genders, and the impact of belonging to multiple minoritized groups or identities.

Autism has changed from childhood to lifespan

Having once been considered a childhood disorder, an improved understanding of Autism has led to a focus on understanding its impact across the entire lifespan, with a particular emphasis being placed on the study of developmental trajectories. This change is exemplified in the terminology used to describe Autism, having first been labelled as ‘Infantile Autism’ in the diagnostic manuals, to now being described as ‘Autism Spectrum Disorder’ in DSM-5-TR ¹. Today, many clinics will have separate child and adult Autism diagnostic services, with adult services frequently assessing adults of all ages for Autism, including older people.

However, despite this shift, the historical impact of Autism being under the purview of child psychiatrists and paediatricians has resulted in little being known about Autism in midlife and older age. In 2022, it was reported that less than 1% of all Autism publications focused on those in midlife and older age ⁶. A recent study of UK primary healthcare records found that only 0.1% of the UK population over 50 years old had an Autism diagnosis, compared to 3% of people aged 10-14 ². The authors estimated that up to 1.2 million people in the UK may be autistic but undiagnosed (600,000 aged over 50). This means that approximately 9 out of 10 autistic people over the age of 50 are undiagnosed (or misdiagnosed) and their support needs may not be fully met. As older age is associated with a broad range of physical, mental, and lifestyle changes, ensuring older autistic people receive support is of paramount importance.

There are many important topics about Autism across the lifespan, particularly in midlife and older age, that need more research attention. For example, a small but growing literature on autistic experiences of menopause indicates that many people struggle with sensory and emotional changes, which can lead to periods of crisis ³²⁻³⁴. Social isolation is also a key topic; Autistic people often have limited social networks throughout their lives and the risk of isolation increases with age. Some studies have identified that many autistic people in midlife and older age have very few social connections, which is associated with poor mental health and quality of life ³⁵. And finally, autistic people appear to be at a higher risk of dementia, which is a leading cause of death in the general population ³⁶. There are many more areas to be explored, and the ageing in Autism research field is rapidly changing.

Autism has changed from categorical to dimensional

The original conceptualisation of Autism described it as a distinct entity, qualitatively different from typical development or other neurodevelopmental conditions. However, the defining behavioural traits and characteristics of Autism are found to be continuously distributed in the general population ³⁷⁻³⁹. Individual differences in autistic traits also seem to be influenced by the same genetic factors that influence diagnosed autism ⁴⁰. As a result, Autism is now viewed as being at the extreme of this dimension of traits and characteristics.

Measuring autistic traits (with self- or parent-report questionnaires such as the Autism Spectrum Quotient)

can be very useful. Autistic traits are assessed clinically as part of the screening and diagnostic process for Autism, and they are also often examined within research. The study of those with ‘high’ autistic traits or the ‘broad autism phenotype’ has become commonplace, and can be particularly effective when studying historically under-diagnosed populations (e.g., women, older people). However, it is important to note that these traits are not exclusive to Autism, and people may score highly on trait measures due to other factors, e.g., social anxiety, trauma, and other conditions ⁴¹. Further research is required to examine whether the cognitive and behavioural features of Autism are found to be reliably associated with autistic traits.

Autism has changed from ‘one Autism’ to many ‘Autisms’

Despite an early acknowledgement of the heterogeneity of Autism (including by Kanner in the 1940s), clinicians and scientists have searched in vain for a single causal factor ⁴². It is currently believed that the heterogeneity in the Autism spectrum likely reflects many different aetiologies ⁴³. This may be why rather little progress has been made in understanding the biology of Autism. Efforts are currently being made to understand this heterogeneity, and the search for stratification biomarkers to discover Autism subgroups for ‘personalised’ interventions is already underway ⁴⁴.

In addition to the ‘many Autisms’ hypothesis, it has been suggested that Autism is best understood as a ‘composite’ condition, reflecting a combination of genetic influences, cognitive facets, and behavioural dimensions.⁴⁵ This ‘fractionated triad’ hypothesis suggests that different aspects of Autism (social, communication, rigid/repetitive behaviour) have separable underpinnings in the same individual, i.e., different genetic factors that influence different traits and characteristics of Autism, resulting in heterogenous presentations ⁴⁵. Studies using twin data have found that the social and non-social aspects of Autism only have a modest genetic overlap ⁴², with many people only having social, but not non-social, autistic traits, or vice-versa. This may result in important clinical considerations. For example, some research suggests that autistic people who primarily have problems with social communication (with their inflexibility problems being managed) may be at a heightened risk of behavioural and emotional difficulties ⁴⁶, while those primarily with inflexibility problems (with their social impairment being managed) may be at increased risk of restrictive or disordered eating ⁴⁷. Thus, understanding the symptom profile of an individual is important to ensure their support needs are being met.

Autism has changed from ‘pure’ to complex

Historically, many diagnoses were viewed in a hierarchal way, with some conditions overwriting others. As a result of this, someone with an Autism diagnosis could not also be diagnosed with other conditions, such as anxiety and ADHD. However, under the new diagnostic guidelines that debuted in DSM-5 ¹, Autism could, for the first time, be diagnosed alongside other conditions.

A change was made in recognition of the fact that Autism is rarely seen in a ‘pure’ form, and most commonly is part of a range of co-occurring conditions. Estimates suggest that 70% of autistic children ¹⁰ and 80% of autistic adults ^{7,48} have at least one other mental health condition. The individual rates of different psychiatric diagnoses were examined in a recent meta-analysis ⁴⁹. It was identified that 33% of autistic people also had an ADHD diagnosis, 23% had an anxiety disorder, 13% had a sleep–wake disorder, 12% had a depressive disorder, 10% had an obsessive–compulsive disorder, 10% had disruptive/impulse-control/conduct disorders, 5% had a schizophrenia spectrum disorder, and 5% had bipolar disorder. And similar rates have been found in autistic people of all ages ^{7,9,48,50}.

Much research has considered why autistic people have such high rates of psychiatric conditions. Many factors have been suggested, such as a shared aetiology (i.e., the genetic ‘*p* factor’ which influences the risk of all psychiatric conditions ⁵¹), environmental factors (e.g., bullying and social exclusion resulting in stress, culminating in poorer mental health), communication differences (e.g., reduced help-seeking resulting in poorer health), or cognitive style (e.g., differences with processing negative events resulting in increased rates of post-traumatic stress). Despite this, a single answer has yet to be found.

However, it is widely accepted that these high rates of co-occurring psychiatric conditions are problematic. For many autistic people, it is not their Autism per se that makes life hard for them, it is their co-occurring conditions which lead to them having difficulties and, in some cases, having periods of crisis. Worryingly, rates of suicide are notably high in autistic populations ^{52–54}, with some studies reporting a 7-fold increased risk for suicide ^{52,55}. In the general population, more men than women die by suicide, but the opposite has

been found in autistic populations. These findings emphasise the need for interventions to be designed for autistic people and research to take transdiagnostic approaches to ensure that the needs of autistic children and adults are being met.

Autism has changed from ‘developmental disorder’ to neurodivergence

Over the past decade, there has been a notable shift in how Autism is perceived by autistic people, clinicians and researchers, and the wider general population ⁵⁶. Rather than being solely viewed through a medical model, Autism is now often viewed through a social model of disability. The notion that Autism is exclusively a disorder defined purely by deficits has been challenged. Instead, many now view Autism as a neurological difference – or ‘neurodivergence’ – with difficulties and disabilities arising based on the poor fit with the demands of living in a neurotypical world.

Additionally, Autism is also often viewed as an identity, with many autistic people becoming advocates, campaigners, or Autism researchers. This shift towards a neurodiversity perspective has been driven by autistic voices. It is notable that the first – and highly influential – autistic autobiography, by Temple Grandin, was published in 1986, and today there are hundreds of first-person accounts. However, the diverse views of the autistic community have not always come together in harmonious ways, and unfortunately the views of autistic self-advocates sometimes clash with the views of parents advocating on behalf of their autistic offspring with significant support needs. Additionally, some neurotypical Autism researchers have clashed with the autistic community, particularly those interested in basic science (e.g., genetics), where the direct benefit of their research for autistic people is not always clearly explained or justified.

A step towards bringing these diverse voices together is participatory research methods, where autistic people, their families, and other stakeholders are included in the design, implementation, and interpretation of research. Historically, most research has been scientist/clinician-led, but now co-produced research is becoming very common ⁵⁷. However, it is important for researchers and clinicians to ensure that *all* voices are heard, including those not actively involved in Autism research and those with intellectual/language disabilities who may require others to advocate on their behalf ⁵⁸.

Opportunities for the future

The world of Autism is rapidly evolving. We have seen great changes to how Autism is conceptualised and understood over the past several decades, with the past few years, in particular, ushering in a new age of co-production and embedding autistic voices into research and clinical practice. But, as with any research area, there are continued opportunities for the future. For example, we are in an age of ‘big data science’, and Autism research has moved from predominately small clinic samples to large cohorts of autistic people of all ages. By utilising these large data sets - for example, national health care records and registries – it will be possible to explore individual differences in Autism, by following an autistic person through their lifespan. This will provide insight into developmental trajectories and how support needs change throughout a person’s life.

There is also a great opportunity for Autism research to become globally represented, as highlighted by the January 2024 special issue of ‘*Autism*’ journal focusing on ‘Autism Research in a Global Context’ ^{59,60}. This special issue emphasises that, today, much research comes from high-income Western countries (particularly Europe and North America) where the population is predominately white. However, most autistic people live in low and middle-income countries, in Asia, Africa, and South America ⁶¹. Societal factors often play an important role in how autistic people are identified and supported, and the models of Autism created in high-income countries may not be fit for purpose in other regions of the world with differing demographics and societal structures ⁶².

Conclusions

Since first being included as its own standalone condition in the DSM-III in 1980, we have seen Autism evolve from being a narrow, rare, mostly male, childhood, discrete and ‘pure’ developmental disorder to now in DSM-5-TR being a wide, common, inclusive of all genders, lifespan, dimensional and complex form of neurodivergence. It is almost a certainty that the field of Autism research and clinical practice will continue

to evolve as our understanding continues to develop, placing us in an exciting place in time to see what becomes of Autism in the near and distant future.

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Recommended Further Reading

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