Autism is now 73 years old and our clinical and scientific understanding of it remains largely disorder-focused, reflected in the name of the diagnostic category in DSM-5: Autism Spectrum Disorder (ASD). Autism is not alone in DSM-5 in being called a disorder. Since DSM-1 in 1952, when there were 106 disorders listed, there has been a steady increase, and when DSM-5 was published in 2013, the number had reached 300. It is unlikely that DSM really ‘carves nature at its joints’, as Plato recommended our best classificatory and explanatory theories should, if we can keep adding or subtracting diagnostic categories each time a new edition of DSM is published. Recall how homosexuality was classified as a disorder in DSM-I and DSM-II, until civil rights protests succeeded in having it declassified from DSM-III in 1980, on the grounds that it is just a natural example of the diversity of sexual orientations that exist in any population.

There has been much debate about whether the DSM-5 characterisation of ASD represented progress (Lai, Lombardo, Chakrabarti, & Baron-Cohen, 2013), but this editorial focuses on the question of whether autism is properly characterised as a disorder and whether the neurodiversity framework should be embraced.

I will use the term ‘autism’ as shorthand for what others refer to either as Autism Spectrum Disorder (ASD) or Autism Spectrum Conditions (ASC), while acknowledging that autism contains huge heterogeneity (from individuals with no functional language and severe developmental delay to what in Europe is still called Asperger syndrome, where individuals have at least average if not above average IQ, and no history of language delay). But what individuals on this autism spectrum all share – relative to age- and IQ-matched individuals without autism – are the social communication difficulties, difficulties with cognitive empathy or theory of mind, the difficulties adjusting to unexpected change, a love of repetition or ‘need for sameness’, unusually narrow interests, and sensory hyper- and hyposensitivities. Autism is also associated with cognitive strengths, notably in domains such as excellent attention to detail, excellent memory for detail and a strong drive to detect patterns (or ‘systemising’) (Baron-Cohen, 2006).

So is autism a disorder? One definition of the term disorder is a ‘lack of order or intelligible pattern’, while another definition of disorder is ‘randomness’. Neither of these seems appropriate to describe autism. The medical definition of the term ‘disorder’ is distinct from the definition of a ‘disease’. The term ‘disease’ is used when a disorder can be ascribed to a specific causal mechanism (that is dysfunctional). In contrast, the term ‘disorder’ is used when the causal mechanism is unknown. Expressed differently, the term ‘disorder’ is intended to simply be descriptive. But it could rightly be argued that the term ‘disorder’ is not just descriptive. The term ‘disorder’ implies the natural order has gone awry and that the individual’s underlying cognition and neurobiology is dysfunctional is some way – even if for environmental reasons. But when we examine the cognition and biology of autism, arguably what we see is not evidence of dysfunction but rather evidence of difference (Lai, Lombardo, & Baron-Cohen, 2014).

Let us start at the genetic level: While it is true that in about 12% of people with autism, rare inherited or de novo single nucleotide variants (SNVs) or copy number variants (CNVs) are associated with autism, almost 50% of the genetic associations with autism involve inherited common variants such as single nucleotide polymorphisms (SNPs) (Huguet, Benabou, & Bourgeron, 2016). See Figure 1. SNPs arise in more than one form in the population and these different forms are not signs of either disorder or dysfunction, and simply reflect difference or natural variation. When rare genetic mutations do arise, they tend to cause much more than just autism, but given that most people with autism do not have such mutations, where is the evidence at the genetic level that autism necessarily involves disorder or dysfunction?

Let us move to the neural level: Here we see plenty of evidence that the autistic brain is different to the typical brain, but one would be hard pushed to pinpoint an example of brain disorder in autism. At the structural level, we see that some regions of the autistic brain (such as the amygdala, at least in childhood) are larger, and others (such as the posterior section of the corpus callosum) are smaller. But differences in size do not add up to evidence for disorder. Volumetric studies of the whole brain in autism sometimes also show early brain overgrowth, but all of these are signs that the autistic brain is developing differently to the typical brain – not direct evidence for neuropathology. Postmortem studies of
the autistic brain reveal a greater number of neurons, for example in the frontal lobe, suggesting that there may be reduced apoptosis in autism, but even this – if we are being careful with our language – is evidence for a different pattern of brain development, rather than a necessary sign of disorder.

At the level of the single neuron, Tang et al. (2014) found an increased number of dendritic spines in autistic neurons, compared to the typical neuron, again reflecting possible reduced apoptosis in autism, and resulting not only in more neurons but also more synapses and connections between neurons in the autistic brain. Some now regard autism as best characterised as a difference in neuronal connectivity – the brain is wired differently – but again no clear evidence of dysfunction or disorder.

Functional MRI (fMRI) studies of the autistic brain at times show less brain activity during some tasks (e.g. in the left inferior frontal gyrus during a ‘theory of mind’ task such as the ‘Reading the Mind in the Eyes’ test), a task on which people with autism show lower scores (Baron-Cohen et al., 1999); or less brain activity during an analytical perception task such as the Embedded Figures Test, a task on which people with autism show higher scores (Ring et al., 1999). Such results may be interpreted as a neural confirmation of social-cognitive deficits, or as support for the view that examples of cognitive talent in autism (e.g. in attention to detail) show a more efficient pattern of brain activity.

Yet other fMRI studies show greater brain activity in autism during an auditory perception test (Samson et al., 2011), which can be interpreted as a neural confirmation of the sensory hypersensitivity that people with autism report when questionnaire studies are conducted. Overall, the fMRI evidence can be interpreted in terms of difference (the autistic brain is processing detail differently), and disability (e.g. the well-established theory of mind difficulties in autism are accompanied by neural differences), but again, no clear sign of disorder.

What about at the behavioural and cognitive levels? Here, studies of people with autism also show both differences, and signs of disability. Differences include finding that young toddlers with autism may look for longer at nonsocial stimuli than at social stimuli, an atypical pattern of attention reflecting what they find of most interest, but not a clear sign of disorder (Pierce, Conant, Hazin, Stoner, & Desmond, 2011). People with autism may show their best performance on IQ tests on the Block Design subtest, perhaps reflecting their strong aptitude for disassembling complex information into its component parts, a key prerequisite for hypersystemising, or understanding how a system works (Baron-Cohen, 2006). These examples of cognitive talent are clearly incompatible with the notion of autism as a disorder. Aspects of social cognition reflect areas of disability in autism, and if a person is not showing any sign of disability, he or she would not warrant a diagnosis. But the language of disability is very different to the language of disorder. Disability requires societal support, acceptance of difference and diversity, and societal ‘reasonable adjustment’, while disorder is usually taken to require cure or treatment. These are very different frameworks.

The relatively new concept of neurodiversity may be very valuable in resolving these findings. The term is thought to have first been used by Judy Singer, an Australian social scientist who herself has autism, and first appeared in print in an article by the journalist Harvey Blume in the Atlantic (3 September 1998). When I first wrote about whether Asperger syndrome should be considered a disorder in the year 2000, in the millennium edition of Development and Psychopathology, I had not heard of the term,
but I was essentially asking the same question. Many in the autism community adopt the neurodiversity framework, coining the term ‘neurotypical’ to describe the majority brain. Steve Silberman’s terrific book Neurotribes is a kind of manifesto for the neurodiversity movement, encouraging us to recognise autism as an example of diversity in the set of all possible brains, none of which is ‘normal’ and all of which are simply different.

The notion of neurodiversity is highly compatible with the civil rights plea for minorities to be accepted with respect and dignity, and not be pathologised. Left-handers are an example of neurodiversity in a majority right-handed world, and left-handers used to be seen as a pathological condition that needed correction. In the same way, the concept of neurodiversity challenges whether people with autism should automatically be ‘treated’ and ‘normalised’. Of course, parents deserve the freedom of choice to give their child early interventions, just as parents of all children make choices of how to promote their child’s potential. Equally, adults with autism deserve the freedom of choice to try different treatments or interventions on offer. But such freedoms are very different to the old default assumption that autism is a disorder that needs to be eradicated, prevented or cured.

Neurodiversity as a term is related to the much more familiar concept of biodiversity, and we now recognise the importance of respecting our environment, with the rich diversity of life forms that inhabit it. In many ways, the concept of neurodiversity is just the next step in this more respectful way of thinking about our planet and our communities.

The neurodiversity framework is not without its critics. Some will object that a child with autism who has epilepsy is not an example of neurodiversity but rather he or she has a disorder. And they are right. Epilepsy is a sign of brain dysfunction and causes disorder (fits) and should be medically treated. But epilepsy, while commonly co-occurring with autism, is not autism itself. Others may say that a child who has language delay or severe learning difficulties is not an example of neurodiversity but has a disorder, and I would support their demand for treatments to maximise the child’s potential in both language and learning. But again, although commonly co-occurring, these are not autism itself.

A final objection to the notion of neurodiversity and a defence of the term ‘disorder’ is that the latter signals the severity of a person’s condition. To counter this objection, the term ‘disability’ can also signal severity, but without the stigma of the term ‘disorder’. Many feel that the term ‘disability’ is softer, and that the term ‘disorder’ is quite hard-hitting. Moreover, while the concepts of disability and neurodiversity are not incompatible, the concepts of disorder and neurodiversity are incompatible. This is because we all have areas of strength or difficulty and some cognitive profiles are adaptive in certain environmental niches but manifest as a disability in others. If someone is tone-deaf, that is only a disability in a situation where the person is expected to sing. Expanding on the quote attributed to Einstein, a fish will appear as having a disability if required to climb a tree. And to expand on the quote attributed to a person with autism, ‘we are fresh water fish in salt water. Put us in fresh water and we are fine. Put us in salt water and we struggle to survive’.

There is little or no challenge to the use of the term ‘disorder’ for conditions such as major depression or severe anxiety, or anorexia or psychosis, because these result in the person no longer being able to function, in any environment. Expressed differently, in these conditions, there is plenty of evidence of ‘dysfunction’. But the case for not applying the term ‘disorder’ to autism is that, in an autism-friendly environment, the person can function not just well, but sometimes even at a higher level than a typical individual. People have made the case for including other neurodevelopmental conditions and ‘phenotypes’, such as ADHD, dyslexia, dyspraxia, dyscalculia or synaesthesia, within the framework of neurodiversity.

This helps to nail down when the terms ‘disorder’, ‘disability’, ‘difference’ and ‘disease’ should be used. ‘Disorder’ should be used when there is nothing positive about the condition, or when despite trying different environmental modifications, the person is still unable to function. ‘Disease’ should be used when the biomedical mechanistic cause of a disorder becomes known, perhaps through medical testing or through scientific research. ‘Disability’ should be used when the person falls below an average level of functioning in one or more psychological or physical functions, and where the individual needs support or intervention. ‘Difference’ should be used when the person is simply atypical, for biological reasons, relative to a population norm, but where this difference does not necessarily affect functioning or well-being. Using these definitions, we should remain open-minded that some forms of autism are properly thought of a disorder, and others not, given the heterogeneity that exists within this diagnostic category.

There are several messages from the concept of neurodiversity. First, there is no single way for a brain to be normal, as there are many ways for the brain to be wired up and reach adulthood. Second, we need more ethical, nonstigmatising language and concepts for thinking about people who are different and/or who have disabilities. Third, we need a framework that does not pathologise and focus disproportionately on what the person struggles with, and instead takes a more balanced view, to give equal attention to what the person can do. And finally, genetic or other kinds of biological variation are intrinsic to the person’s identity, their sense of
self and personhood, which seen through a human rights lens, should be given equal respect alongside any other form of diversity, such as gender.

It will be important to see how the concept of neurodiversity is applied to the 300 diagnoses currently all called ‘disorders’ in DSM-5, and if it revolutionises both the science and the practice of psychiatry.

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