AUTISM SPECTRUM DISORDER

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"It takes a village to raise a child. It takes a child with autism to raise the consciousness of the village."

Coach Elaine Hall, 2005

1. Introduction

The term "*autism*" comes from the Greek word "*autos"*, meaning "*self"*, used to describe conditions of social withdrawal – or the isolated self. Autistic Spectrum Disorder (ASD) is a neurodevelopmental syndrome in medicine. The aim of this research is to develop a sound scientific understanding of ASD from a critical and humanistic perspective. We investigated the symptoms and the potential causes of ASD, as well as critically analysed its current diagnostic method. We also discussed the social implications of ASD.

2. Symptoms

As much as autism is a spectrum disorder, which means that the symptoms and severity are varied across the spectrum, there are still some manifestations of ASD that autistic individuals with different severity share. These symptoms can be categorised into social interaction, patterned behaviour, and sensory processing.

2.1 Social Interaction

Autistic individuals often have difficulty understanding the social use of language (Northumberland County Council Communication Support Services, UK. 2004). Even for high functioning autistic individuals whose intellectual capacity is at or above the average level, they seem to not understand language use like jokes or sarcasm and need direct instructions instead. Individuals on this spectrum also manifest a lack in ability to express their feelings and empathise with others because they often cannot recognise emotions (Sebastian Gaigg, 2014). This, however, is not tantamount to saying that they do not have feelings. They are capable of experiencing a range of emotions despite their difficulty in showing or identifying them. This inability often results in their deficits in social-emotional reciprocity. For example, some of them are unable to maintain a back and forth conversation while others suffer from a complete lack of facial expressions and nonverbal communication (American Psychiatric Association, 2013).

2.2 Patterned Behaviour

Many autistic children manifest stereotyped behaviour which includes repetitive body movement, speech and movement of objects (Autism Research Institute). Some examples include flapping hands, rubbing one's skin against another object and idiosyncratic phrases. People with ASD may also establish certain routines for themselves and insist on following these routines. Their insistence on sameness could lead to extreme distress when they experience a change. Some individuals experience anxiety attack when they enter a new environment, which could lead to an autistic meltdown, i.e, an explosion of emotions.

2.3 Sensory Processing

The sensory processing of an autistic brain is not synchronised with real time (Judy Endow, 2015). While they might be hypersensitive to some sensory stimuli, they appear to be hyposensitive to others. Due to their hypersensitivity to certain sensations, some autistic individuals show strong emotive reactions to a particular stimulus. For example, some autistic children are extremely afraid of the sound of a bell. Furthermore, it is found that some autistic individuals have problem comprehending real-time auditory information, which is not associated with their intellectual capacity. For instance, they may hear one say the word "book" clearly, but do not understand what it means at the moment. In addition, due to their low-level multisensory integration (Marco et al., 2011), too much information could lead to sensory overload for these individuals, resulting in a distortion of senses and an autistic meltdown as a coping mechanism for the brain.

3. Etiology of Autism Spectrum Disorder

Since the term "autism" was first used by a Swiss psychiatrist Eugen Bleuler (1857 – 1939) to refer to one group of symptoms related to schizophrenia, scientists embarked on the journey to understand Autism Spectrum Disorder. Over the years, various theories have been proposed to explain the causes of ASD but none are completely successful. Although correlations are often observed, today scientists still cannot understand the etiology of ASD. In this section, we will discuss three possible theories of the causes of ASD and evidence to support them.

3.1 Abnormal Connectivity

In recent years, scientists are studying how brains of autistic individuals are different from brains of neuro-typical people. Scientists reach a consensus that it is the unusual coordination of different regions in the brain that affects the brain functioning of people with ASD. Their studies are supported by evidence from neuroimaging tools such as functional Magnetic Resonance Imaging (fMRI) and Diffusion Tensor Imaging (DTI). Abnormal connectivity hypotheses include, but are not limited to, the following three.

- Underconnectivity hypothesis

  With the help of fMRI, scientists found that in the context of specific tasks, inter-regional (particularly the frontal-posterior) connectivity in the brain is disrupted in autism, hence the autistic individuals have difficulty forming patterns of thought, especially when the computational demand is large (Just et al., 2012). This hypothesis also posits that the underconnectivity can be attributed to the lower communication bandwidth among cortical areas, particularly between frontal and posterior areas, in people with ASD than typical participants (Just et al., 2012).

Figure 1. (Left) Schematic depiction of typical systems connectivity. (Right) Schematic depiction of lower bandwidth between frontal and posterior cortical centers in autism.

- Local overconnectivity hypothesis

Although long-distance connectivity in ASD is lower than normal level, studies show that local functional connectivity atypically increases in adolescents with ASD in temporo-occipital regions bilaterally (Keown et al., 2013). This local overconnectivity hypothesis is supported by evidence from fMRI and graph theory. As local connectivity is positively correlated with Autism Diagnostic Observation Schedule (Lord et al., 2000), the extend of overconnectivity in particular brain regions is linked to severity of ASD.

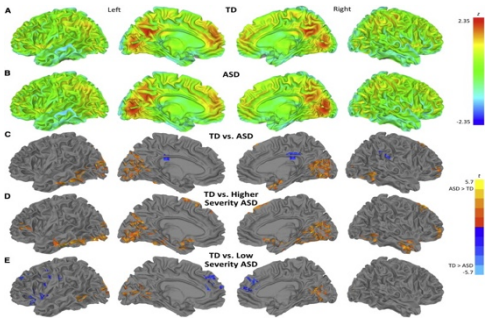

Figure 2. Within- and Between-Group Maps for Local Connection Density

(A–E) Surface renderings of local connectivity density for TD (A) and ASD groups (B). Greater *Z* scores correspond to brain regions with high connectivity (red scale). Clusters of significant group differences in local degrees ($p < 0.05$; corrected [corr.]) for entire ASD cohort (C) as well as for higher-severity (D) and low-severity (E) ASD subgroups in comparison to the TD group (warm colors: ASD > TD; cool colors TD > ASD).

- Distortion of spontaneous connectivity hypothesis

This hypothesis proposes that weaker inter-hemispheric synchronisation is the cause of ASD. For example, poor understanding of social use of language, a typical symptom of ASD, can be explained by weak synchronization in IFG and/or STG, two areas commonly associated with language processing (Dinstein et al., 2011). Notably, the neuroimaging tool used to support this hypothesis is resting-state fMRI performed when toddlers are sleeping. This might be a useful tool for early screening of ASD if this hypothesis is developed further and reliable biomarkers are identified.

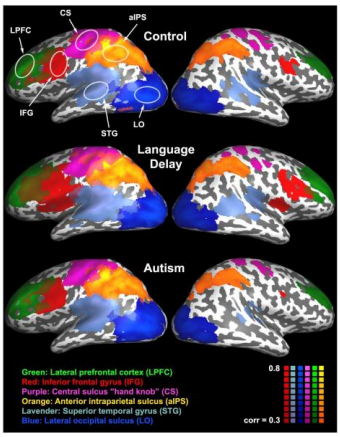

Figure 3. Correlation maps averaged across toddlers from the typically-developing (top), language delay (middle), and autism (bottom) groups.

3.2 Genetics

ASD has a strong genetic basis. Identical twin studies put autism's heritability in a range between 36% and 95.7%, which means that 36% and 95.7% of the differences between autistic and non-autistic individuals is due to genetics (Muhle et al., 2004). Some examples of identified autism susceptibility genes include: protein LRRN3 and LRRTM3 gene (Sousa et al., 2010), CHD9 gene (Wang et al., 2009), and mutations in GABRB3 gene (Buxbaum et al., 2002). Scientists recently identified 18 new ASD-risk genes by analysing whole-genome sequence of 5205 people, about half of whom have autism (Yuen et al., 2017). However, there is no convincing evidence of any single gene or multigene interaction that can be the causative determinants of ASD, and it is unclear whether ASD is explained more by multiple gene

interactions or by rare mutations with major effects (Abrahams & Geschwind, 2008).

3.3 Neurological disorders

Some scientists have identified similarities between neurological disorders and autism. Studies have shown that children with certain neurological diseases, such as Rett syndrome and Fragile X syndrome, do display characteristics that are similar to autism (Chaste et al., 2012). It was also discovered that these neurological diseases have something in common — they disrupt the production and degradation of molecules in the brain, which are responsible for controlling the quantity and quality of synapses, the region allowing nerve cells to transmit signals to each other (Lüscher & Isaac, 2009).

Rett syndrome and autism do share many common features. For Rett syndrome, it involves the mutation of MECP2 gene, responsible for the development and maintenance of neurons. A mutated MECP2 gene causes neurons to be small and simplified dendrites with reduced and immature dendritic spines to be formed (Percy, 2011).

Fragile X syndrome, as the name suggests, is an X-linked disorder. For individuals with Fragile X syndrome, more than one third display signs of autism. Fragile X syndrome results from the blocked expression of the fragile X mental retardation protein (FMRP). A lack of expression of this gene causes dendrites to grow wildly and contain long and thin spines, which do not mature to an adult morphology (Comery et al., 1997).

All these conditions are not isolated as individuals with autism might have more than one neurological disorders. As much is not known about these diseases yet, it cannot be concluded that these neurological diseases are the sole cause of autism. However, what can be said is that these diseases lead to an increased risk for autism, as many who suffer from this disease are diagnosed with autism too.

3.4 Prenatal Environmental Factors

Research increasingly suggests that ASD arises from a combination of genetic susceptibility and environmental triggers, in particular, factors in the prenatal environment. Pregnancy-related exposures have been the focus of epidemiologic of ASD for over 40 years. One prenatal risk factor is the infection of viruses during pregnancy. Very recently a group of Norwegian scientists investigated the relationship between active infection of Herpes simplex virus type 2 (HSV-2) in early pregnancy of woman and the probability of autistic children and found that the presence of high levels of anti-HSV-2 antibodies at midpregnancy doubles the risk of ASD in boys, proving that infection of virus does increase chances of autism in children (Mahic et al., 2017).

3.5 Social Construction Theory

The social construction theory argues that ASD does not exist as an objective entity but a socially

constructed label to distinguish a group of people from others. The diagnosed group, the proponents suggest, inhabit the identities that have been ascribed to them, and promote their sense of well-being by resisting or appropriating autistic ascriptions (Nadesan, 2011). Scientific evidence would have little value to support such theory. Mainly, sceptics argue from the point of philosophy of science that the boundary between normal and abnormal state is subjectively and arbitrarily drawn. In other words, there is no objective standard to distinguish the "autistic" from the rest. Social construction theory challenges the diagnosis of ASD, which will be further discussed in the next section.

4. Diagnosis of Autism Spectrum Disorder

The severity of ASD varies widely among individuals, making the diagnosis difficult. Currently, the diagnosis of ASD relies heavily if not solely on the Diagnostic and Statistical Manual of Mental Disorders (DSM-5), published by the American Psychiatric Association (APA) in 2013. DSM-5 broadened the definition of autism and condensed autism, Asperger syndrome and pervasive developmental disorder-not otherwise specified (PDD-NOS) into a single diagnosis - ASD. The diagnostic criteria for ASD include "impaired social and communication skills" and "restricted, patterned behaviour" (Mayo Clinic).

The use of DSM-5 grants diagnosis of ASD universality as physicians across the world are using the same set of standards to diagnose ASD. Nevertheless, we must be aware of the limitations of using this guideline. Physicians look for various symptoms (see "Section 2. Symptoms") before making clinical judgement, such judgement is only based on observable evidence and human-invented evaluation tool. There is no biomarker to be used to clinically diagnose ASD yet. Besides, the diagnostic criteria for ASD in DSM have changed over the years, so the basis of diagnosis varies. One interesting fact regarding this is that although the prevalence of ASD has risen (in 2014, the Centers for Disease Control and Prevention estimates that 1 in 68 children in US has ASD as compared to 1 in 88 reported in 2008), we cannot conclude that ASD are becoming more common in reality. The increased awareness of ASD and easier access to quality medical care definitely account for the rise in prevalence. The importance of public awareness will be discussed in the next section.

5. Importance of Understanding Autism Spectrum Disorder

Understanding the causes of autism is crucial for coming up with therapies. Given that many hypotheses coexist to explain the causes of ASD, we need to scrutinise each one closely and eliminate those that lack solid scientific evidence. For example, scientists initially suggested that vaccines could overwhelm a child's immune system thereby predisposing them to ASD. Subsequent scientific studies refuted the causal relationship between vaccinations and ASD (Taylor et al., 2014). The earlier we narrow down the exact causes of autism are, the earlier effective screening and preventive measures can be taken to prevent potential full-fledged development of autism (Parizad Bilimoria).

Furthermore, due to the interdisciplinary nature of neuroscience, researchers have recently observed surprising overlapping and converging goals and challenges of autism research and affective computing. A collaboration between two fields could lead to several mutually beneficial outcomes — from developing new tools to assist people with autism in understanding and operating in the socioemotional world around them, to developing new computational models and theories that will enable technology to be modified to provide an overall better socioemotional experience to all people who use it (El Kaliouby, Picard, Baron-Cohen, 2006).

More awareness of this disorder also helps in removing the stigma associated with ASD. According to a recent study by Drexel University, 58 percent of young adults with autism are unemployed. However, their ability to focus over a long period of time can actually give them an edge in certain jobs such as spotting anomalies from large bodies of information (Bourree, 2016). By understanding more about ASD, this disorder can be turned into an asset rather than a shame that people on this spectrum are stuck with. Furthermore, with more people being able to identify autistic individuals in their social circle and understand why they behave in certain manner, these autistic individuals can better integrate into society.

6. Learning Points

In this research, we have examined autism from multidimensional perspectives, including the genetics and the neurology of autism, as well as its close link to society. From its symptoms to its potential causes, our understanding of autism has developed beyond a simplistic one. Systematic research was conducted to summarise the existing prominent theories that explain the causes for autism. Among them, the genetics theory has the most solid scientific basis. The social construct theory, on the other hand, challenges the current model, arguing that autism is essentially a social label constructed by the mainstream society. This theory is often overlooked by many researchers and practitioners in the study of autism. Nonetheless, it provides us with critical insights and reflections on how individuals with autism are perceived in society and how their identities can be shaped by these perceptions. We have also evaluated the limitations of the current diagnosis guidelines and discussed potential epistemic problems in identifying ASD cases. To sum up, we feel that while striving for a comprehensive understanding of autism spectrum disorder, we should also bear in mind that the end-goal of research in this field is to improve the quality of life for autistic individuals and their families, be it the development of effective early screening, intervention and treatment methods or efforts in destigmatising autism spectrum disorder.

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Text

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