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Autism's anatomy

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2 | What is this thing called autism?⁴

To act, it is necessary at least to localize. For example, how do we take action against an earthquake or hurricane? The impetus behind every ontological theory of disease undoubtedly derives from therapeutic need. When we see in every sick man someone whose being has been augmented or diminished, we are somewhat reassured, for what a man has lost can be restored to him, and what has entered him can also leave. (Canguilhem, 1966/1991: 39)

How does one find a law for irregular phenomena? – this is the fundamental problem of medical thinking. (Fleck, 1927/1986: 39)

Abstract

Currently, autism is a widespread and diverse neurodevelopmental disorder that includes both severely impaired and institutionalized patients and the fairly geeky but brilliant university professor. Despite its heterogeneity, autism is often presented as a distinct nosological entity with a unifying autism essence. This chapter argues that the common belief about the ontological status of autism is that autism constitutes a *natural kind*. There are, however, two major difficulties with a natural kind approach in autism research. First, how can we continue to speak about autism as a distinct disease while the condition is marked by such a sheer diversity of symptoms, traits, biological markers and cognitive profiles? And second, recent historical works on autism illustrate that there is something fundamentally social and historical about how autism is defined, diagnosed and treated. I argue that the dominant natural kind approach in autism research is problematic, as autism can only be understood in relation to ideas about what kind of behavior is deviant and in need of correction or

⁴ This chapter has been published as Verhoeff B (2012) What is this thing called autism? A critical analysis of the tenacious search for autism's essence. *Biosocieties* 7(4): 410-432.

support. Furthermore, locating and maintaining autism within the biological realm of the individual obscures an array of social, cultural and psychological issues in understanding the contemporary phenomenon we call autism.

Introduction

Autism has many faces. Besides autistic disorder, we are now familiar with Asperger's syndrome and 'high functioning' autism, currently all united under the banner of the autism spectrum. As autism researchers and clinicians increasingly highlight the dimensional nature and complex behavioral, biological and neurocognitive heterogeneity of autism (see, for example, Happé and Ronald, 2008), autism has become a broad and diverse disorder covering both completely mute and unresponsive individuals, and socially awkward geniuses. At the same time, despite battles over its causes and how it should be classified in the *Diagnostic and Statistical Manual of Mental Disorders (DSM-5)*,⁵ the idea of autism as a distinct nosological entity with a particular essential core deficit remains pervasive. Many influential clinicians and autism researchers emphasize the recognizability and distinctiveness of autism: '[T]here is little dispute that in many ways autism is one of the better examples of a "disorder" that is distinctive and does not just shade off into normalcy' (Volkmar, 1998: 45) and 'Autism is a recognizable entity' (Frith, 1989: 15). Leo Kanner himself, autism's intellectual father, was also convinced that he delineated a distinct entity: '[W]e can state unreservedly that, whether or not autism is viewed as a member of the species schizophrenia, it does represent a "definitely distinguishable disease." This disease, specific – that is, unique, unduplicated – in its manifestations, can be explored per se' (Kanner, 1965: 418).

In this chapter, I illustrate that in the expanding field of autism research, the common – if sometimes tacit – belief about the ontological status of autism is that autism constitutes a *natural kind*. In spite of the disputable philosophical status of the very notion of a natural kind (see Hacking, 2007a), a natural kind perspective matters in scientific practice. Natural kinds make good objects of

⁵ *DSM-5* was published in 2013 by the American Psychiatric Association, after the publication of this article in *Biosocieties*. Different Work Groups, consisting of approximately ten to fifteen experts in a particular field, were responsible for formulating specific criteria for each disorder.

scientific discovery, are related with the search for mechanistic explanations, and allow for inductive generalizations.⁶ Current autism research fits this scientific image strikingly well. It is guided and regulated by the depiction of autism as a *bona fide* scientific and physical object that can be discovered and identified with systematic biomedical and neuroscientific investigation. Autism is imagined to exist as an objective entity independent of its embodiment in particular individuals. Conceptualized in such a way, the idea of autism as a distinct disease with a specific etiology, pathophysiology, course and treatment seems inescapable.

However, I argue that a natural kind approach in autism research is thorny in at least two ways. First, is a problem widely acknowledged among autism researchers and clinicians, that is, the issue of heterogeneity: how can we continue to speak about autism as a distinct disease while the condition is marked by such sheer diversity of symptoms, traits, biological markers and cognitive profiles? Natural kinds⁷ – classic examples being *water* and *gold* – typically refer to classes of things that are homogeneous and have clear boundaries. Moreover, the individual members of a natural kind must share some underlying structure or property that characterizes the kind in all possible cultures, historical periods and worlds in which it could exist (Dupré, 1981). Natural kinds seem to carve up the world at naturally privileged joints. However, despite an impressive list of genes, anatomical abnormalities and other biological markers associated with autism (see, for example, Anagnostou and Taylor, 2011; Lichtenstein et al., 2010), a diagnosis of autism is still based

⁶ Inductive generalizations are ‘formulated on the basis of observed instances of a category and yet purport to license inferences about unobserved instances as well’ (Samuels, 2009: 51). On observing 100 glasses of water that all boil at 100° Celsius it is reasonable to expect that all glasses of water boil at 100° Celsius. Such inferences about unobserved characteristics are reliable if the observed characteristics of a category covary not by accident, but because of some common factor that explains the co-variation of important characteristics. This is part of what defines a natural kind: that the members share a similar structure that determines a number of properties equally shared by the members of the natural kind.

⁷ Different accounts of natural kindhood have been associated with many different classes of things, such as chemical elements and biological species. Some accounts are very stringent and others are more permissive (see Boyd, 1991), but it is not my intention to get drawn into metaphysical disputes about what exactly defines a natural kind. I will use the term rather loosely to illustrate how a certain ontological assumption shapes and regulates scientific practices.

on behavioral characteristics and there is a broad consensus that unlike in Down syndrome or Huntington's chorea, people with autism do not possess a unitary and essential causal property that determines all observable characteristics of autism (Happé et al., 2006).

An obvious question that arises is how the idea of autism as a distinct natural entity remains pervasive while empirical evidence against an essential structure underlying autism accumulates? An answer to this question might be found by analyzing social and political forces that locate and maintain autism within the biological realm of the individual rather than between the individual and the requirements of the environment. Social and political structures involved in the production of suffering and disability remain unchallenged as long as autism is a biological thing to be identified with microscopes and MRI scans. Notwithstanding the relevance and potential explanatory value of 'constructionist accounts in terms of medical imperialism, medicalization of social problems, energetic proselytising by parents and support groups, the egregious power of the drug companies with their disease awareness campaigns ...' (Rose, 2009: 79), it will not be the primary theme of this chapter.

Rather, in the first out of two parts of this chapter, I discuss the way in which some central developments and discussions in the field of autism are both conditional upon and further legitimize the particular understanding of autism as a reputable natural entity. After that, the way in which the supposed essence of autism is currently conceptualized will be discussed and I give an account of the actual strategies by which autism researchers attempt to deal with autism's heterogeneity. These strategies are related to different research programs and some of the debates around the proposed changes to the autism category in *DSM-5*.

A second difficulty with a natural kind approach in autism research emerges with a more philosophical, historical and sociological viewpoint on autism. In the growing field of critical autism studies there are extensive discussions about defining autism as a disease (Ortega, 2009), and about the emergence of autistic cultures, subjectivities and (self-)advocacy movements (Silverman, 2008). In particular, some recent historical works on autism by Nadesan (2005), Eyal et al. (2010) and Silverman (2011) forcefully illustrate that there is something fundamentally social and historical about how autism is defined, diagnosed and treated. They all argue that autism's emergence, historical transformations and fluctuating boundaries reflect social, cultural and political processes. Drawing

amply on Ian Hacking's notion of 'interactive kinds,' Eyal et al. (2010: 209) for example argue that 'classifying and naming autism set in motion processes that act on the phenomena classified and possibly change them in ways that, in turn, react back on the classification, leading to its revision'.

With a particular emphasis on Hacking's notion of an interactive kind, the second part of this chapter explores the extent to which autism's historical variability threatens a natural kind approach in autism research. Despite the importance that the idea of an interactive kind may have for explaining historical changeability of autism and for drawing attention to the power and social effects of labeling humans, I suggest that this idea will not suffice to criticize a dominant natural kind approach in autism research. Nevertheless, supported by more historical, socio-cultural and philosophical analysis, I argue that autism can only be understood in relation to ideas about what kind of behavior is unacceptable, deviant, and in need of correction or support. Autism cannot avoid being related to a cultural norm of a social, empathic and engaged individual, and any account of autism begins with a need to demarcate, locate and treat particular discontents and impairments that have appeared. Therefore, the idea of an essential core or a natural autism entity that is waiting to be identified is misguided, and the corresponding dominant neuroscientific approach to autism obscures an array of social, cultural and psychological issues important in understanding the phenomenon we call autism.

The analysis in this chapter is based largely on texts. Manuals, articles, reviews and books from multiple disciplines provide the foundation of the arguments. I also conducted interviews with some of the members and one ex-member – Fred Volkmar – of the *DSM-5* neurodevelopmental disorders (ND) Work Group that was responsible for the formation of criteria and diagnostic categories of autism and related disorders.⁸ The interviewees were selected on the basis of their expertise and authority regarding the topic of a subsequent article that will specifically cover the emergence of the *autism spectrum disorder* (ASD) category and the disappearance of Asperger's disorder as a specific disease entity in *DSM-5*. Thus, whilst this chapter does not explore these interviews in depth, I will occasionally use quotes from these interviews to illustrate some of my arguments.

⁸ See: <http://www.dsm5.org/MeetUs/Pages/Neurodevelopmental%20Disorders.aspx>, accessed 10 April 2012.

Part I: A natural entity

An autism epidemic?

In 2009, the Center for Disease Control and Prevention (CDC, 2009) published a study that estimated that the prevalence of autism in the United States had increased from 4 per 10,000 in 1989, to 66 per 10,000 in 2002, to 90 per 10,000 in 2006. That is from 1 in 2,500 children to 1 in 110 children in less than twenty years. The CDC added that other recent population based studies documented even higher prevalence rates of autism of >1 per cent of children in areas of Japan, Sweden, the United Kingdom and the United States (for example, Baird et al., 2006; Kogan et al., 2009). The substantial and steady rise in children diagnosed with autism has been of scientific concern. Multiple review articles and meta-analyses have tried to explain what caused this tremendous increase in people diagnosed with autism (see, for example, Fombonne, 2009; Waterhouse, 2008; Wing and Potter, 2002).

At this point, I am not particularly concerned with the specific explanations given for this rise nor with adding a new one, but deliberation on the potential increase of autism requires a certain notion of autism. Although different autism researchers might slightly differ in the explanations they give for the rise in autism cases, there is a striking similarity in the way the problem is posed and possible explanations are presented. The articles by Eric Fombonne, psychiatrist and autism epidemiologist, are exemplary. He reframes the problem of the rise in autism cases into a deceptively simple question with two possible outcomes. Either there is a true increase in the incidence of autism, or there is no true increase in the incidence of autism and only an increase in recognition of it.

Fombonne (2005: 292) argues that while the rates of people diagnosed with autism have gone up, ‘this increase most likely represents changes in the concepts, definitions, service availability and awareness of autistic-spectrum disorders in both the lay and professional public’. He concludes that current evidence ‘does not support the hypothesis of a secular increase in the incidence of autism’ (ibid.). Fombonne’s idea of a secular or true increase in autism assumes an idea of a true autism not at the level of concepts, definitions or diagnostic criteria, but at some ‘underlying’ material level. Concepts, definitions and criteria of autism may change, but true autism is a stable and material, but still largely unidentified, disease. Francesca Happé – prominent autism

researcher and member of the *DSM-5* ND Work Group – makes a similar argument: ‘there has been a big change in diagnostic criteria, a broadening, the introduction of Asperger’s syndrome, a widening of understanding, better diagnostic services, better services to follow diagnosis ... whether they are sufficient to explain the [rising] numbers or whether there is a real increase, I think nobody knows’.⁹

Current hypotheses about such a real increase in autism point at several causal factors including antibiotics, environmental pollutants, *de novo* genetic mutations, preservatives in vaccines and other neurotoxins.¹⁰ While none of these factors have been affirmed as a cause for the increase in autism rates, they are among the possible candidates for causing a ‘genuine’ autism increase. ‘A true risk’, Michael Rutter (2005: 2) argues, ‘due to some, as yet to be identified, environmental risk factor cannot be ruled out’. On this view, changing ideas about autism cannot explain a true increase in autism, for the reason that autism is imagined as an entity (or multiple entities) existing in individuals prior to and independent of our ideas about it.

A critique of thinking about autism as a natural entity does not imply that the rising number of people diagnosed could not for instance be caused by environmental pollution or *de novo* genetic mutations. In addition, claims and worries about an autism ‘epidemic’ do not depend exclusively on conceiving of autism as a natural phenomenon. However, the assumption of autism as a natural entity limits the scope of explanations, solutions and interventions in discussions and research on the possibility of an autism epidemic. Social and cultural forces, that are discussed in the second part of this chapter, such as shifting social norms and new ways of engaging with suffering and diversity remain out of sight in a narrow investigation of a ‘true’ increase in autism in biological terms. A good example of a sociological analysis of the more general rise in psychiatric disease incidences is given by Rose (2006), who discusses contemporary ‘demands for disease recognition, moves to screening, fears of risk and hopes for prevention’ as forces related to lower thresholds ‘at which individuals are defined, and define themselves, as suitable cases for treatment’ (p. 481). Such an analysis does not deny an involvement of biological or genetic

⁹ From an interview with Francesca Happé in London on 14 December 2010.

¹⁰ See for the controversial but extremely influential MMR vaccine hypothesis in the retracted article by Wakefield et al. (1998).

factors, but broadens the perspective on current rising numbers of a variety of diseases from hypertension to obesity and ADHD.

Retrospective diagnosing

Another indication of the natural kind assumption in autism research comes from the currently popular practice of retrospective diagnosing (see also Chapter 3). While the first official case descriptions of autism date back to Leo Kanner's 1943 study, many autism experts have been occupied with demonstrating that there are striking cases of autism in history before Kanner introduced the syndrome. Uta Frith, another leading autism researcher, describes various cases, of which the case of Victor, 'the wild boy from Aveyron' is among the most prominent. At the turn of the nineteenth century, Victor was found living wild in the woods in South-Central France. The French physician Itard published detailed accounts of his behavior. Frith finds in Itard's early accounts clear evidence for serious impairment in reciprocal social interactions and stereotypical behavior. She assumes that Victor must have been autistic and concludes that 'autism is not a modern phenomenon, even though it has only been recognized in modern times' (Frith, 1989: 16).

Other historical accounts of feral children, eccentric geniuses and religious figures have proven productive for retrospectively diagnosing autism. To name a few, there are *The Blessed Fools of Old Russia* (Challis and Dewey, 1974), the extraordinary case of Hugh Blair of Borgue in eighteenth-century Scotland (Houston and Frith, 2000) and – according to Michael Fitzgerald's (2005) endless list – Isaac Newton, Ludwig Wittgenstein and Albert Einstein.¹¹ These are just a few examples that for many provide convincing evidence that autism is indeed not a contemporary phenomenon, but something that can be recognized in different cultural and historical contexts. Frith, Fitzgerald and others explicitly distinguish the existence of labels from the existence of distinct diseases. According to Frith, retrospective diagnosing helps to 'distil those features that are the essence of the disorder beyond our immediate time and cultural context' (Frith, 1989: 17). She tries to find what she calls 'the unchanging core of autism' (Houston and Frith, 2000: 4). It is this assumed transhistorical, essential core of autism that is being recognized in the many

¹¹ Michael Fitzgerald, a Professor of Child Psychiatry, just about specializes in diagnosing historical legends with autism. He argues for a link between autism and a kind of pure and innate artistic and philosophical brilliance (2005).

recent examples of peculiar historical figures, and these examples strengthen – for autism experts and authorities, but also for clinicians, lay people, patients and their families – the idea of autism as a natural entity. Though, as we shall see, what this actually means is far from settled.

Autism as a brain disorder

Recent articles and reviews on autism usually start with stating that autism is a ‘neuropsychiatric condition,’ a ‘brain disorder’ or a ‘neurodevelopmental disorder’ (see also Chapter 4 and, for example, Happé et al., 2006; Volkmar and Pauls, 2003). Autism is considered to be ‘among the most heritable of all mental disorders’ (Lichtenstein et al., 2010): recent reviews estimate the heritability of autism to be more than 90 percent (Freitag et al., 2010; Losh et al., 2008). The emphasis on genetics, brain activity and neurotransmitters is not specific to autism but part of a broader neuroscientific shift in psychiatry that hopes – amongst other things – to carve psychiatric classifications at their ‘natural joints’ (Hyman, 2007). Social, cultural and historical conditions of possibility for this neuroscientific shift have been explored by theorists in great detail (see, for example, Rose, 2007), but fall outside the scope of this chapter.

Against earlier psychogenic theories of autism, of which the ‘refrigerator mother’ theory¹² commonly attributed to Bruno Bettelheim (1967) is among the most remarkable, the biological nature of autism is currently treated as a given starting point for solving ‘the autism puzzle’ (Schaaf and Zoghbi, 2011). Thinking about autism is far removed from unconscious desires of mothers, family dynamics and defense mechanisms of the child. Notwithstanding an increasing interest in ‘gene-environment interactions’ and epigenetics (Rutter, 2011), what there is to be explained can and should ultimately be explained at the material and mechanical level of the brain in terms of genetic polymorphisms and synaptic connectivity. This biological depiction of autism goes beyond arguing that the etiology of autism involves biological factors. Indeed, it is not only about what causes autism, it is about what autism

¹² The popularization of this term ‘refrigerator mother’ is often attributed to Bruno Bettelheim and his widely read book *The Empty Fortress* (Bettelheim, 1967). The psychoanalytically founded claim in this book is that the precipitating factor for autism is the unconscious wish, of particularly the mother, that the child should not exist (see Chapter 5). It were, however, Kanner (1949) and Eisenberg and Kanner (1956) who originally mentioned ‘a frosty atmosphere’ and ‘emotional refrigeration’ in relation to the environment and the parents of their autistic patients.

fundamentally is: a disorder of the brain, identifiable and discoverable in this particular organ at the biological and mechanical level commonly studied by the biomedical and neurosciences. The self-evident supposition of autism as a brain disorder is another facet in thinking about autism as a natural entity.

Autism today

Ubiquitous heterogeneity

What does this natural entity called autism look like? As I already mentioned in the introduction, present-day autism researchers and clinicians increasingly emphasize the heterogeneity of autism. Lord and Jones (2012: 491) recently stated that ‘the most significant scientific challenge to the concept of autism as one “disease” or even “diseases” is the heterogeneity of the genetic findings’, and, in addition, Lord (2011: 166) pointed out that ‘anyone who has met more than one person with an ASD is stuck by the differences between these individuals’. *DSM-IV* (APA, 1994) diagnostic criteria for autism are based on the characteristic autism triad of impairments that consists of impairment in social interaction, communication and impairment due to restricted behavioral patterns. With the introduction of the idea of a broad autism spectrum, Wing and Gould (1979) already highlighted that this characteristic ‘triad of autism impairments’ could occur at varying levels of severity in different individuals with autism.

Impairment in social interaction, for instance, can be expressed as a total avoidance of social interaction, or it can be expressed as a lack of understanding of social conventions and cues in a person who does show interest in other people and friendships. Impairments in communication can range from a complete lack of speech to fluent speech with particularities in intonation, pitch and rhythm, and restricted interests and repetitive behavior can be as diverse as spinning a coin for hours, or having an obsessive interest in train tables or fantasy books. Apart from the possible variety in visible signs and symptoms, the course of autism also varies from children who seem to develop normal for two years and then show a decline, to children who show an abnormal development from birth on. Some children improve with age, others have a stable course and some get worse (Willemsen-Swinkels and Buitelaar, 2002). Furthermore, autism has been associated with numerous other conditions such

as epilepsy, intellectual disabilities, sleep disorders, disruptive behaviors, anxiety, depression, hyperactivity and attention difficulties.

In their article with the telling title ‘Time to give up on a single explanation for autism’, Happé and colleagues (2006) not only argue that there is no single cause for all the core features of autism, but they also present empirical data – contra Wing – of behavioral fractionation of the communication impairment, the social impairment and the rigid and repetitive behaviors. In a population-based study, they found ‘modest-to-low correlations between autistic-like behavioral traits in the three core areas’ (ibid.: 1218). The clustering of the three symptom domains turned out to be not as strong as was previously thought, and many children only have symptoms – in varying levels of severity – from one or two symptom domains. This finding adds another level of heterogeneity to the spectrum idea of autism, now that there is variation and fractionation along at least three partly independent dimensions of impairment.

Happé and colleagues furthermore claim that it is useless to search for a monolithic genetic or neuroanatomical explanation for the three core aspects of autism as a whole. Family and twin studies of the genetic structure of the triad of impairments and neuroimaging research all suggest an equal fractionation of underlying genetic abnormalities and neural substrates. For future research, they recommend neuroimaging and molecular genetic studies to focus on specific symptom domains, instead of the so far unsuccessful ‘search for genes [and neural substrates] “for autism” as a whole’ (2006: 1219).

Have genetic studies indeed been unsuccessful in their search for autism genes? The first advances in identifying genes associated with autism came from studying syndromic autism (autism in combination with other syndromes that cause congenital malformations or dysmorphic features). Identified single-gene syndromes, such as fragile X syndrome, PTEN macrocephaly syndrome, Rett syndrome and tuberous sclerosis, are assumed to account for 5 to 7 percent of all autism cases (Miles, 2011). Another 5 percent of autism cases have been associated with genetic metabolic disorders, such as phenylketonuria, creatine deficiency and mitochondrial disorders. For the remaining – nonsyndromic – cases, recent whole genome studies on autism (Levy et al., 2011; Sanders et al., 2011) further affirmed the genetic heterogeneity in autism. These studies argue that there are numerous genetic mutations – *de novo* and transmitted – associated with autism that are extremely rare. The number of genes associated with autism may be a couple of hundreds or more, of which

the most common mutations were found in just over 1 percent of the children with autism, and not exclusively in children with autism (Schaaf and Zoghbi, 2011).

Autism's unifying essence

With this amount of diversity at clinical and biological levels, and with the absence of a clear boundary between autism and normality, what is it that unifies all the cases of autism other than the diagnostic criteria? In what sense and at what level is autism still a distinct natural entity? For many prominent autism experts, autism remains to be understood as a highly recognizable disease entity with a unifying fundamental nature. Happé and Ronald (2008: 299) for instance argue that their idea of the fractionable triad is not an attack on the validity of the diagnosis of autism since 'it is quite compatible to assert that ASD results when a number of independent impairments co-occur, and to assert that the resulting mix has a special quality, distinct prognosis and response to intervention'.

In a paper on classifying autism in the *DSM-5*, Wing, Gould, and Gillberg (2011: 769) state that 'the fundamental problems underlying all autistic conditions and the Triad of Impairments, is absence or impairment of the social instinct present from birth ... [w]e hope that research work into the behavioral neurology of the social instinct will be carried out in the near future'. Happé adds to this that '[autism] is one of the most recognizable syndromes. It is a clearly recognizable syndrome when you see it ... the receptionist who works in the clinic spots the autism as they come in the door'.¹³ Its recognizability, she argues, lies in the overlapping essential deficit in social cognition: '[autism] is a disorder of social cognition ... social problems are the real core'. On this issue Susan Swedo – chair of the *DSM-5* ND Work Group – argues similarly: 'yes, there is something that is essentially autism and the essential is a delay or failure of the development of social cognition ... autism is a core deficit, it is a pathway that did not develop'.¹⁴

Various distinct cognitive theories have been suggested as fundamental in comprehending all the problems and impairments people with autism are faced with. A defective theory of mind (Baron-Cohen, 1995), weak central coherence (Happé and Frith, 2006), impaired executive functioning (Hill, 2004), and

¹³ From an interview with Francesca Happé in London on 14 December 2010.

¹⁴ From an interview with Susan Swedo in New York on 29 October 2010.

dominant systemizing capacities compared to empathizing capacities (Baron-Cohen, 2002) are the familiar cognitive theories that have all been proposed as the essential uniform defect in autism. However, none of these theories have succeeded in explaining all the problems in autism. Happé and colleagues (Happé et al., 2006: 1219) admit the ‘failure to find a single cognitive account for the three core features of autism’. In a recent review, Chevallier et al. (2012) have a new attempt by proposing ‘the social motivation theory of autism’. They argue that autism can be seen as an ‘extreme case of early-onset diminished social motivation’ that explains a range of autism characteristics, ‘including cascading effects on the development of mature social cognitive skills’ (p. 237). Furthermore, they argue that ‘these deficits appear to be rooted in biological disruptions of the orbitofrontal-striatal-amygdala circuitry, as well as in dysregulation of certain neuropeptides and neurotransmitters’ (ibid.).

For many cognitive psychologists, clinicians and neuroscientists researching social cognition, and – as we will see in the next section – to a lesser degree for molecular geneticists, an essential nature beneath the diversity of apparent signs and symptoms is still assumed to unite the multiplicity of autism. Expressed in terms of a deficit in social cognition, social intuition or social motivation, and imagined somewhere in the brain, identifying autism is widely presented as a complex scientific challenge, often metaphorically referred to as ‘the autism puzzle’ or ‘the autism enigma’.¹⁵ This rhetoric of puzzles and enigmas is reliant upon the idea of autism as one or more distinct disorders that can be discovered, but it also has a number of other effects: it is effective in soliciting funding and broad public support, it serves as a magnet for researchers invested in resolving mysteries, and legitimizes the already highly specialized and costly searches taking place at increasingly complex neurobiological levels. As the assumed natural entity of autism cannot be identified at a concrete genetic or neuroanatomical level, the attempts to reveal autism’s mystery continue at even more minute and detailed molecular and epigenetic levels (Rutter, 2011). Preliminary ideas about how this natural entity is envisioned and how this relates to attempts of autism researchers to deal with heterogeneity issues are discussed in the next section.

¹⁵ A recent special issue of *Nature* called ‘The autism enigma’ attempts to sort fact from fiction in autism research (*Nature* 2011, volume 479).

Dealing with variety and autism in *DSM-5*

Lumping

Attempts to deal with heterogeneity and to identify the essential underlying nature of autism can be roughly divided into two familiar types of strategies: *lumping* and *splitting* (see McKusick, 1969). Lumping refers to the attempt to identify a single unifying underlying process or structure that offers an all-encompassing account of autism. The numerous and diverse genetic variations and associated neuroanatomical structures in different autism patients obviously pose a problem for a lumping strategy. However, a possible solution is provided by the idea of a more homogeneous mechanism or structure at a different level – sometimes referred to as a ‘final common pathway’ – that connects multiple distinct etiological and pathological processes with overt behavioral autism characteristics.

Geschwind and Levitt (2007: 103) suggest a potential unifying model for autism in which particular areas of the brain that normally connect to the frontal lobe are partially disconnected during development. Their concept of developmental disconnection, they argue, ‘can accommodate the specific neurobehavioral features that are observed in autism, their emergence during development, and the heterogeneity of autism etiology, behaviors and cognition’. More specifically, recent genome studies by Gilman et al. (2011) and Levy et al. (2011) suggest that the many different genes associated with autism could play a functional role, via the molecules they express, in a larger biological network that is related to the regulation and formation of synapses and neural development. Corresponding to a more general network-based approach to human diseases (see Barabasi et al., 2011), this larger biological network responsible for neuronal motility, axon guidance and synaptogenesis, could be disturbed by many different molecules expressed by different genes, but leading to similar deficits of neural development. These specific deficits in neural development are in turn thought to underlie the typical symptoms identified as autistic. In a similar vein, Sakai et al. (2011) developed a protein interaction network that ‘provides a framework for identifying causes of idiopathic autism and for understanding molecular pathways that underpin both syndromic and idiopathic ASDs’, and Peça and Feng (2012) illustrate how multiple families of genes involved in tuberous sclerosis, fragile X syndrome, Angelman syndrome and non-syndromic ASD, are functionally united by common cellular pathways

involved in the organization of glutamatergic synapses. Hence, a common neurodevelopmental abnormality at a complex biological level could still be the glue that holds the overall idea of autism together.

Nevertheless, a lumping strategy still needs to deal with the diversity of clinical presentations of autism. What makes it reasonable to assume that the diverse manifestations are the result of a common underlying problem? The *DSM-5* ND Work Group clearly opts for a lumping approach in the proposal to fold Asperger's disorder into a single ASD category.¹⁶ As stated by one of its prominent members, the aim in proposing the new ASD category is to 'recognize the *essential* shared features of the autism spectrum' (Happé, 2011: 541, emphasis added). At the same time the Work Group members recognize a 'vast heterogeneity within this spectrum' (ibid.: 540). They attempt to deal with it by specifying individual levels of symptom severity and by assigning nonspecific dimensional descriptors (co-existing conditions), such as intellectual or language difficulties, mood disorders and motor or sleep problems, to an ASD diagnosis.

Although core deficits – in social cognition and intuition – unite all ASD patients, their diagnosis will be individualized by describing overall levels of impairment from co-existing difficulties to account for the unique problems of each ASD patient. Particularly, levels of intellectual functioning and language competence are considered to have a major influence on autism's manifestations and on clinical decision-making. Lord and Jones (2012: 493) affirm that the 'level of expressive language is a crucial dimension in the diagnosis of ASD that needs to be considered separately in its own right, and as it affects ASD symptoms'. Other commonly associated conditions like hyperactivity, aggression, eating abnormalities, anxiety and sleep disorders, are equally not part of the core deficits in autism, but are thought to have a huge impact on how autism develops, impairs and is expressed. Therefore, these conditions are able to account for much of the heterogeneity seen in ASD. The idea of *dimensions*, both within the autism core in terms of severity of symptoms and as referring to co-existing conditions and impairments, is crucial to the reorganization of autism in *DSM-5*. It is able to unite extreme clinical heterogeneity with the idea of one single all-encompassing autism category.

¹⁶ See: <http://www.dsm5.org/proposedrevision/Pages/Default.aspx>, accessed 10 April 2012.

Splitting

Splitting autism into subgroups is another approach that attempts to deal with the problem of heterogeneity. Folstein (2006), for instance, argues for two ‘true’ autism phenotypes, Kanner’s autism and Asperger’s disorder. She regards Kanner’s autism as characterized by a child’s severe inability to relate himself to other people and situations, language deficits and an intolerance of change of any type, such as changes in a certain routine or furniture arrangement. Asperger’s disorder is a (genetically) related milder variant, but Folstein argues that both autism diagnoses should exclude a number of other conditions. Children with profound mental retardation, children with dysmorphic facial features or specific genetic conditions, such as tuberous sclerosis or Rett syndrome and children who have suffered certain kinds of severe encephalitis at an early age should be excluded from the two autism categories. Folstein argues that while these children might exhibit autism-like behavior, they are etiologically very heterogeneous and need to be considered separately from autism in neurobiological studies.

Waterhouse (2008: 283) suggests that we should accept that all the behavioral and biological variation in autism cannot be encompassed by any single theory and there ‘must be phenotypic and genotypic subgroups that have not yet been discovered’. This idea of multiple autism subgroups has led geneticists to a research approach that attempts to identify genes and regions on the genome that are associated with more homogeneous subgroups within the autism spectrum. By splitting the autism spectrum into smaller phenotypes, for instance of children with autism who also suffer from epilepsy, language delay, or extreme resistance to change, geneticists were able to identify new regions on the genome associated with only a subgroup of autism cases (see, for example, Molloy et al., 2005; Abrahams and Geschwind, 2008). However, apart from some single-gene syndromes such as fragile X and Rett syndrome, no convincing and meaningful subgroups have been identified so far. Furthermore, it is still largely unclear how the earlier mentioned whole genome studies (Levy et al., 2011; Sanders et al., 2011), that suggest the involvement of hundreds of rare genetic mutations, relate to the breakup of the autism spectrum into distinct (genetic) disorders. Future ASD research, Geschwind (2011) argues, should focus on identifying subtypes of ASD based on clusters of (interacting) genes and converging molecular pathways that relate to specific deficits in brain circuits that in turn relate to distinct autism phenotypes.

For (molecular) geneticists and neuroscientists, functional genomics, epigenetics and systems biology are among the new hopes in a new phase of solving the autism puzzle. Whether they are more committed to the idea of multiple disease entities (splitting) or to the idea of one broad autism entity (lumping), they all share a faith in discovering objective disease categories. The idea of splitting up autism into a number of genetically or neurobiologically defined disorders is not in conflict with a natural kind approach in autism research, as it is equally driven by the idea of discovering and delineating unified natural entities by means of systematic neuroscientific investigation. However, in contrast with most cognitive psychologists, neuroscientists researching social cognition and clinicians, (molecular) geneticists are generally not that much committed to the idea of autism's unifying deficit in social cognition or intuition. Since their focus is on genes, molecules and neural circuits in defining and categorizing distinct disorders that underlie the superficial autism features, the idea of a unifying essential deficit in social cognition becomes less urgent. But, as I have mentioned above, how autism should be split up on the basis of neurobiological data, what the corresponding (autism) phenotypes will be and how they might be related to each other is still largely undetermined (see Geschwind, 2011; Berg and Geschwind, 2012).

***DSM-5* debates**

For as long as autism is not yet carved at its natural joints and the search for distinct biological mechanisms continues, the behavior-based DSM classification system remains the main point of departure both for clinical practice and for fundamental neurobiological research. The recent proposal to create one ASD category for the *DSM-5* has aroused heated debates and high emotions. Particularly the incorporation of Asperger's disorder into ASD has encountered forceful resistance from psychiatrists, researchers, patients, patients associations and other stakeholders (see, for example, Ghaziuddin, 2011). However, the Work Group argues that 'there is little evidence to support the current diagnostic distinction between Asperger disorder and high-functioning autism' (Happé, 2011: 541).

Among the opponents of the ND Work Group's decisions is autism expert Fred Volkmar. He resigned from the Work Group in 2009 based on his objections to the single ASD category and on his worry that less children will qualify for extra support and therapeutic services. Volkmar, supporting a

splitting approach, argues that Asperger's disorder should be in *DSM-5* as a separate category 'both for research and clinical purposes'. He suggests that 'there is a big difference with autism ... [Asperger's children] have different needs, they are more verbal and look for social relationships, you can use that as a medium for intervention, and they often have a different profile of strengths and weaknesses'.¹⁷ From a research perspective the Asperger's disorder category is important because of 'the potential differences from autism in neuropsychology, and I also think genetics is probably stronger in Asperger's'. The problem with *DSM-5* is that 'they get rid of all the distinctions [...] If you could really find a homogeneous subgroup, you can find genes, but if you just mess them all up as a spectrum we are not able to do that'. Furthermore, Volkmar worries that 'a lot of the people with Asperger's disorder will probably lose their diagnosis altogether [...] because if you look at the proposed criteria, they have put up the threshold. That is one of those funny things, you think spectrum means broader, but it is actually narrower'.¹⁸

As a senior author, Volkmar recently backed up his earlier worries in a provocative study (McPartland et al., 2012) that mapped the new *DSM-5* criteria for ASD onto a dataset of child and adult patients collected more than 20 years ago for the field trial of *DSM-IV*. They conclude that almost 40 percent of individuals currently diagnosed with an ASD, mostly individuals with Asperger's disorder and PDD-NOS, will no longer meet criteria for an ASD in *DSM-5*. The proposed changes, the authors predict, 'could exert detrimental effects on service eligibility' (p. 382). These alarming conclusions generated substantial media attention and parental concerns with *DSM-5*, because of the fear that a significant proportion of individuals with a current autism diagnosis will be excluded from medical, educational and social services.

In a commentary in the same journal issue, the entire ND Work Group fires back and argues that the McPartland et al. study justifies 'neither alarming headlines nor dramatic conclusions' (Swedo et al., 2012: 347). So far, the Work Group has been careful to stress that 'no individual currently diagnosed with Asperger's disorder or PDD-NOS who needs support will lose that support' (Happé, 2011: 541), and they believe the data used in McPartland's analysis 'have too many inherent limitations to assess definitely the criteria proposed ... and do not support such dramatic conclusions' (Swedo et al., 2012: 347). The

¹⁷ From an interview with Fred Volkmar in New Haven CT 27 October 2010.

¹⁸ Ibid.

issue of service eligibility illustrates a certain tension that arises with the production of a diagnostic manual intended to serve a whole range of different purposes that extend beyond structuring research and clinical practice. The very sensitive topic of eligibility for therapeutic services and support is one important aspect in redefining autism that intermingles with the scientific interest of identifying distinct biological mechanisms. As we will see in the second part of this chapter, such contextual issues pose profound difficulties for a natural kind approach in autism research.

Part II: Historical variability

Recent social studies on the emergence and development of autism suggest that there is something fundamentally social and historically variable about how autism is defined, treated and diagnosed. Silverman (2011: 29) argues that although practitioners may maintain that they have gained the ability to recognize autism on sight in a similar way as one might learn to recognize the distinctive style of a particular artist, ‘much about the diagnostic criteria, practices of identification, modes of treatment, and daily experience of autism has changed, and changed radically’. Indeed, in Kanner’s days, autism was a very rare disorder characterized by a child’s extreme withdrawal and remoteness from affective and communicative contact with other people (Kanner, 1949). These days, as we have seen, autism is a widespread neurodevelopmental disorder with a variety of social and behavioral impairments.

In *The Autism Matrix*, Eyal et al. (2010) explain the historical changes in autism and the exponential rise in the number of autism diagnoses in terms of a process of deinstitutionalization of mental retardation in the mid-1970s, parental activism, a greater availability of services from 1991 onwards ‘when autism was added to the Individuals with Disabilities in Education Act’ (p. 19), and the reorganization of expertise. In a similar vein, Nadesan’s *Constructing Autism* (2005) explores the role of early-twentieth-century child guidance and mental hygiene movements, ‘which together brought childhood into focus as a legitimate sphere of psychiatric inquiry’ (p. 6). She provides a social history of the material institutions, professional identities and cultural values that enabled the emergence and transformation of autism as a psychiatric disorder.

These historical studies approach autism not as a distinct entity in nature that can be discovered, but as something mutable and produced by its socio-historical context. However, a natural kind approach cannot easily be discarded on the basis of autism's historical variability, as autism scientists could argue that human understandings of a (supposed) 'true' nature of autism might improve as science progresses. One might argue, as the retrospective diagnosticians do, that we just get better at recognizing what has always been there. Furthermore, any scientific discovery or progress requires particular social, cultural and institutional conditions of possibility. Probably, some highly 'social' history could be told about the discoveries of the periodic table elements. Nevertheless, I argue that the dominant natural kind approach in autism research is limited. This, however, requires further examination of the precise character of autism's historical variability.

Interactive kinds

In their explanation of the historical variability of autism, Nadesan (2005), Eyal et al. (2010) and Silverman (2011) draw on Ian Hacking's notion of 'interactive kinds'.¹⁹ Silverman (2011: 29) argues that diagnostic categories are interactive, mutable things, 'they make groups of subjects visible and distinct by describing them, but they then set them free to carry on their business, to resist, reshape, and reform that definition through their own actions'. Likewise, for Eyal et al. (2010: 23), the key concept for understanding historical changes in autism is 'looping':

Certain human conditions, says Hacking, are interactive in the sense that the very act of naming, classifying, diagnosing, and assigning them to treatment loops back to modify the condition thus named. [...] We suggest thinking of the autism epidemic, therefore, neither as a naturally

¹⁹ Much of Hacking's work (see Hacking, 2007b) attends to the way in which classifying and describing people brings about a feedback mechanism that changes the kinds of people under study. Individuals react to being classified and as a result of their behavioral changes, the very people and the classifications that are supposed to cover them go through a process of alteration. This feedback mechanism, Hacking argues, results in 'interactive kinds' having histories totally different from the histories of what he calls 'indifferent kinds'. Indifferent kinds, such as tigers and gold, are unaffected by how we classify them.

occurring event, nor as a socially constructed fiction, but as a final spiral in an increasingly widening vortex of looping processes.

Eyal and his colleagues convincingly illustrate this interactive process by arguing that as Leo Kanner and Bernard Rimland – another eminent first generation autism researcher – insisted on the rareness and distinctiveness of autism in the mid-1960s in order to destigmatize the condition, to defeat the by that time dominant psychogenic hypothesis and to remove the burden of guilt from parents, they also gave rise to an active parents’ movement. This meant, they argue, ‘that the diagnosis was becoming less rare, and in due course, less distinctive’ (2010: 210). After that, as therapies emerged, what happened between autistics and therapies ‘began to redefine what autism really was and what it was not’ (ibid.). Therapies identified certain central behaviors and characteristics like head-banging, self injury and extreme social remoteness as treatable and therefore ‘incidental to the syndrome, thus trimming away at its edges and blurring its boundaries’ (ibid.). This, they argue, led to revisions of diagnostic criteria in subsequent DSM editions, and eventually completed the ‘loop’.

Yet can we conclude on the basis of these interactive processes that autism cannot be understood as a discoverable entity in nature, or could it still be argued that these changes reflect a better understanding of what autism really is?²⁰ Or, as Cooper (2004) astutely points out in arguing that interactive kinds can still be natural kinds, could autism be changeable and still be a natural kind in the same sense that bacteria – given that they are natural kinds – mutate as they are affected by antibiotics? Changes in the behavior of people diagnosed with autism due to therapeutic processes or due to reactions on being classified do not contradict a natural kind approach. Although these behavioral changes might be evidence for social, cultural and psychological factors in affecting behavior, they are not evidence for a *necessary* dependence on social or cultural factors for the existence of autism as a distinct category, in a sense that the

²⁰ Contrary to what Eyal et al. seem to suggest, there are several ways to evaluate whether a particular cluster of symptoms makes a more valid category than another cluster of symptoms. Instead of only using an ‘objective’ marker ‘in order to check their validity’ (2010: 19), psychometric techniques such as ‘confirmatory factor analysis’ are used, next to more conventional ‘validators’ such as familial aggregation and precipitating factors. For a recent book on psychiatric taxonomy, see Kendler and Parnas (2012).

existence of the category of domestic animals necessarily depends on how we created it.²¹

Ian Hacking himself (1999; 2007b) has been struggling with autism and this issue of whether natural, or indifferent kinds – as he prefers to call them – and interactive kinds are mutually exclusive. On the one hand, Hacking represents autism as an interactive kind. The doctrine of the refrigerator mother had a severe impact on the family and the person diagnosed with autism and this doctrine and the subsequent changes in the family ‘contributed to a rethinking of what childhood autism is – not because one found out more about it, but because the behavior itself changed’ (Hacking, 1999: 115). A looping effect changed those diagnosed with autism ‘in such a way as to change the very concept of autism’ (Hacking, 2007b: 304).

On the other hand, Hacking leaves open the possibility of autism being – ‘in traditional jargon’ – a natural kind. With the identification of ‘one or more fundamental neurological or biochemical problems’ (Hacking, 1999: 116), then, ‘the more obvious it will seem that we are in the realm of indifferent, “natural” kinds’ (ibid.: 120). After all, for Hacking, it is obvious that ‘there were autistic children before Kanner singled them out’ and ‘if, as is widely supposed, autism is a congenital neurological deficit, then there were certainly autistic children who were dismissed as retarded, feeble-mind, and so on, a long previous litany of dismissive epithets’ (2007b: 304). For Hacking, autism’s interactiveness and historical variability eventually do not threaten a natural kind approach in autism research. As ‘many kinds of mental illness are interactive kinds, and yet are also indifferent kinds’ (1999: 119), autism can still be empirically discovered at fundamental neurological or biochemical levels.

To sum up, the notion of an interactive kind has proven to be a great conceptual tool for analyzing developments in the concept of autism. Particularly, in the works of Eyal et al. (2010), Nadesan (2005) and Silverman (2011), this notion facilitated valuable sociological insights in how ideas about autism emerged and changed and how autism became such a prevalent and much discussed condition. However, the idea of autism as an interactive kind is insufficient to criticize the dominant natural kind approach in autism research. Instead of highlighting autism as an interactive kind, I will now shed light on autism’s variability in a somewhat different way, and challenge Hacking’s hypothetical identification of autism as a natural (indifferent) kind. I argue that

²¹ For the entire argument, see Cooper (2004).

delineating autism and identifying autism at neurobiological levels are fundamentally reliant upon what is considered to be abnormal, harmful and impaired in a child's relation with the world, and thereby, fundamentally reliant upon where we draw the lines. In other words, the 'natural joints' at which autism researchers intent to carve up autism are reliant upon often very implicit commitments to normative expectations of individual children and their behaviors.

The undesirability of autism

As Nadesan (2005: 19) illustrates in her history of the emergence of autism, this event in the 1940s 'must be understood in relation to a matrix of professional and parental practices marking the cultural and economic transitions to the twentieth century'. The formalization of compulsory education and the creation of the child guidance movement led to increased forms of social surveillance over childhood, an increased public concern over 'deviant' children who posed a threat to social stability and an increased demand for more nuanced understandings of childhood pathology. Consequently, understandings of normality and pathology in mental health shifted and community clinics and special schools for children newly recognized as in need of psychiatric evaluation and support emerged. Autism could only emerge as a diagnostic category because, according to Nadesan (*ibid.*: 53), 'it was within these schools and clinics that a new cadre of experts ... encountered a class of children who escaped the increasingly narrow parameters of normality but whose apparent pathologies could not be satisfactorily explained by the extant psychiatric categories'.

Autism, as a problem of sociality, was able to fill the space that came into being by the increasing need to medically structure, treat and regulate childhood deviance. Nadesan's analysis provides an historical example of a widespread consensus among philosophers of psychiatry (see, for example, Bolton, 2008) that how the line between what is normal and what is pathological is drawn depends on social, cultural and individual values and circumstances. There is no plausible way in which this distinction can be made by referring only to biological or statistical measures (Canguilhem, 1966/1991). Even someone like Jerome Wakefield (1992), who is considered to be on the farthest naturalist side concerning mental disorders, acknowledges that a biological dysfunction needs

to be harmful in order to become a disorder, and harm cannot be understood independent of sociocultural circumstances.²²

The evaluative and contextual nature of pathology suggests that the kind ‘mental disorder’ as a whole does not constitute a natural kind, and a quick inference could be that the subclassification of mental disorder into specific mental disorders inherits this lack of natural kindhood. However, many scholars in the philosophy of psychiatry object to such an inference. In defense of a mechanistic model of psychiatric disorders, Kendler et al. (2011: 1147) argue that ‘psychiatric kinds are grounded in common features of the causal structure of the world, not merely imposed upon the world by psychiatrists through their classificatory practices’. Arguing for a natural kind approach to delusions, Samuels (2009) concludes that delusions do not have to be pathological. There is no necessary, but only a contingent connection between pathology on the one hand, and delusions on the other. He states that without some reason to suppose that this connection is a necessary one, the normativity of pathology does not pose a threat to the natural kind thesis in delusions. Cooper (2005: 76) uses an analogy to clarify this point:

...we should think of mental disorders in a way analogous to the way we think about weeds. Weeds are unwanted plants, thus whether a daisy is a weed is at least in part a value-judgement. Still, types of plant that are generally considered to be weeds – daisies, buttercups, stinging nettles – are natural kinds. Similarly, I argue that the claim that schizophrenia is a disorder is in part a value-judgement, but that it may well be the case that schizophrenia and depression are natural kinds.

For Cooper, Samuels and Kendler and colleagues, the general kind ‘mental disorder’ should be considered independent from kinds such as schizophrenia and autism. The pathological aspect of specific mental disorders is not seen as something inherent in psychiatric categories and as a consequence, the possibility of a natural kind approach is repossessed. From a different perspective, a similar position is taken by a group of autism self-advocates – the ‘neurodiversity’ movement – who ‘believe their condition is not a disease to be

²² Wakefield’s notion of *harmful dysfunction* has been influential in the philosophy of psychiatry, but also controversial since his idea of a value free biological dysfunction is highly problematic. For a discussion on this topic see Bolton (2008).

treated and, if possible, cured, but rather a human specificity (like sex or race) that must be equally respected. For them, an atypical neurological “wiring” and not a pathological cognitive organization accounts for their difference’ (Ortega, 2009: 426). A current diagnosis of autism, however, requires a *failure* to develop *appropriate* relationships, a *lack* of seeking to share enjoyment or *impairments* in the use of behaviors that regulate social interaction (APA, 1994). Instead of attributing the impairments and failures to the ‘atypically wired’ autistic individual, the neurodiversity movement locates the source of failure and impairment in the lack of acceptance, respect and societal tolerance for autistic difference.

Nevertheless, irrespective of the possible sources of distress and impairment, and unlike daisies or buttercups, delineating and conceptualizing autism has always been related to the medical commitment to treating suffering, impairment and ‘abnormality’ wherever it occurs. A necessary relation between diagnosing autism and undesirable conditions, often expressed in terms of impairment, disability or distress, can be further illustrated by paying some more attention to diagnostic assessments and instruments. In a clear and accessible book *Autism and Asperger Syndrome: The Facts* (2008), written for a wide audience by autism professor Simon Baron-Cohen, the process of making an autism diagnosis is spelled out. Baron-Cohen explains that, besides standardized diagnostic instruments such as the ADI (*Autism Diagnostic Interview*), a clinician conducting the diagnostic interview needs to ask questions that are evidence for the required social difficulties: ‘Have they found it difficult to make and keep friends? ... Do they show a lack of normal *social awareness*? ... Have they found it hard to understand and respond appropriately to other people’s feelings?’ (Baron-Cohen, 2008: 38). Answers to these types of questions with value-laden terms such as ‘lack of’ or ‘appropriately’ profoundly depend on experiences of impairment or dysfunctioning in the social sphere and on implicit social norms related to making friends, being socially aware and being able to empathize appropriately.

As I mentioned earlier, behavior-based diagnostic manuals – like the DSM – are the main point of departure both for clinical practice and for fundamental neurobiological research, and an official DSM diagnosis for autism even explicitly requires, among other things, qualitative impairments in social interaction and in communication (APA, 1994). In *DSM-III-R* (APA, 1987: 38), one of the criteria for impairment in communication was ‘lack of imaginative

activity ... and lack of interest in stories about imaginary events'. This criterion changed in *DSM-IV* (APA, 1994: 70) into 'lack of varied, spontaneous make-believe play or social imitative play', and in the proposal for *DSM-5*, this criterion changed again into 'difficulties in sharing imaginative play'.²³ These changes in criteria reflect changes in ideas about abnormal and impaired imaginative play in children. In spite of autism's inevitable relation with (variable) ideas about abnormality and impairment in social behavior, Baron-Cohen naively hopes that in the spirit of a natural kind approach 'diagnosis will not depend on the vagaries of a clinical interview or of direct observations of behavior, which invariably includes some subjective elements. Instead, it will be based on a biological marker or set of markers ... measured in the blood or in other bodily tissue or cells' (Baron-Cohen, 2008: 41).

The works of Nadesan (2005), Eyal et al. (2010) and Silverman (2011) provide more examples of how the variable needs of clinicians, parents, researchers and society guided the demarcation and structuring of particular problems with sociality, and how autism emerged and developed in close relation to historically and socially variable ideas about deficiency, abnormal behavior and unmet needs of children. Nadesan (2005: 3), for instance, argues that high-functioning forms of autism and Asperger's disorder emerged 'in the context of new standards for parenting that emerged mid-twentieth century'. Within new cultural and social conditions in the 1960s that led to a shift in 'emphasis on the "psychological" adjustment of personality ... towards an emphasis on "cognitive" fitness of intellect' (p. 109), the popularization of models of intellectual and linguistic development and the ethological idea of 'critical periods' of developments created a need and responsibility for engineering a child's intellectual and emotional development. In combination with a growing public's interest in education and an effort to ensure a child's future success, this led to an increased sensitivity of parents to any 'delays' in a child's cognitive and social development, an increased need for expert advice, new needs and space for medicalizing and pathologizing childhood deviance, and a further expansion of autism's boundaries (Nadesan, 2005).

I suggest that it is a combination of, on the one hand, the context dependent undesirability of autism, and, on the other hand, the context sensitive nature of the problems (with sociality) that autism covers, that explain

²³ See: <http://www.dsm5.org/proposedrevision/Pages/Default.aspx>, accessed 31 August 2012

what makes autism particularly vulnerable to change. Current ideas about autism are relational to some sense of discontent about how a child relates to its environment. This is why, for autism at least, Cooper's weed analogy does not hold good. Shifting ideas of how a child should behave in the social world go hand in hand with shifting scientific ideas on autism, whereas varying ideas on what should be recognized as a weed, will not affect the characteristics of daisies or buttercups. The boundaries of autism are not set by nature, but by the need to frame discontent in a particular way: a way that marks, delineates and converts a capricious category of problems into a suitable case for treatment, a way that facilitates a structured narrative that is able to understand and explain what previously remained uncertain. The needs and discontents of a society regarding how an individual interacts with others, makes friends, initiates chit-chat, seeks to share enjoyment, empathizes and figures out implicit social norms, will be continuously in flux and make autism inevitably hard to pin down. Not a single gene, biological network, neural process or cognitive theory can do anything about the variability of autism.

Conclusion

During autism's relatively short history, it has repeatedly been presented as a distinct nosological entity. All through the many theories that have been suggested and rejected, from refrigerator mothers to vaccine preservatives and genetic mutations, researchers and clinicians have always tried to get a grasp on the true nature of autism.

Discussions on the rising rates of people diagnosed, on how autism should be classified in the *DSM-5*, and other pressing issues like which promising new research directions should be invested in and where treatment and interventions should be directed at are all shaped within a framework that assumes autism to be a natural category. Currently, this 'thing' called autism is imagined at a neurobiological level and autism researchers continue their search at increasingly complex levels of epigenetics, molecular biology and neural networks, to find what they are looking for: the biological boundaries of autism. Despite ubiquitous heterogeneity, the rhetoric of the autism puzzle and the prospect of finding one or more underlying unified mechanisms fuels the

tenacious search for autism's neurobiological nature. This search comprises the central challenge for contemporary autism researchers.

There is, however, a serious concern for a natural kind approach in autism research. No matter how sophisticated and multicausal the underlying mechanisms are presumed to be, autism can only be understood in relation to ideas about what kind of behavior is unacceptable and in need of correction or support. Autism's emergence, the historical transformations and fluctuating boundaries necessarily reflect certain desired standards of a child's connection with the world. Autism has always been related to a cultural norm of a social, communicative, empathic and engaged individual, and recent historical studies on autism show that various changing accounts of autism cannot avoid being related to historically and socially variable needs to demarcate, locate and treat particular discontents and impairments that have apparently appeared. In making this point, I certainly do not argue that autism does not include severely disturbed individuals that immensely suffer, disrupt entire families, and need continuous care. Seriousness or persistency is not at issue here. But I do argue that despite the widely experienced sense of a distinct and unique syndrome, there is no transhistorical essence or 'true' autism core to be revealed in nature, and that there is a remarkably persistent desire to locate suffering, disruption and the requirement for care as a natural phenomenon, rather than implicated in the demands of a social world.

This is not just of theoretical importance. Attempts to carve nature at its joints, to dig up historical cases of autism and to clarify whether there is a 'true' autism epidemic are only intelligible with the presumption that autism can be defined and demarcated in neurobiological terms. Rejecting this presumption by emphasizing the social and cultural elements in delineating autism reveals these attempts to be seriously limited. Furthermore, apart from the enormous costs that are involved in neurobiological research, a natural kind approach in autism research obscures an array of social, cultural and psychological issues important in understanding how the phenomenon we call autism has emerged, developed and become one of the most prevalent mental disorders in children in less than thirty years. What have become the prevailing values and implicit norms in social life and children's behavior? How does modern society deal with diversity and suffering; what does it mean to be a child diagnosed with autism and how does it shape a child's sense of self? What are the social and

political forces that enable to locate and maintain autism within the biological realm of the individual?

These questions – and others – have become relatively unimportant in the scientific search for autism’s natural boundaries at neurobiological levels. In practice, they have been dismissed to the supposedly non-scientific disciplines such as sociology, anthropology and philosophy, and thus designated as fundamentally irrelevant to the work of scientists. However, clinicians and neurobiological researchers urgently need to engage these questions. Acknowledging that autism is not an entity (or multiple entities) contained within an individual and that it does not have a unifying core or fundamental nature, de-inevitabilizes current biomedical perspectives, taxonomies, research purposes and intervention targets. Acknowledging the human and contextual elements in the production of the ‘thing’ called autism makes it possible to renegotiate autism’s boundaries, to put the above-mentioned questions on the agenda in autism research and to better connect with the concerns of those involved. For this purpose, a closer collaboration between the social, medical and neuroscientific disciplines is absolutely indispensable. Only from a multidisciplinary point of view can we properly approach the question whether the current use of the category of autism to delineate, structure and respond to the ‘autistic’ problems so many children and adults are faced with today is effective, useful and ethically warranted.

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