The Autism Matrix

Gil Eyal et al.
THE AUTISM MATRIX
# CONTENTS

**Acknowledgements**  
 viii

**Introduction: The Autism Matrix**  
- Why focus on therapies?  
  9
- The genetics of autism  
  14
- Diagnostic change  
  18
- Between mental illness and mental retardation  
  25

1  
**The Puzzle of Variation in Autism Rates**  
- Diagnostic substitution  
  46
- Supply-side and demand-side explanations for diagnostic substitution  
  49
- Deinstitutionalization as key to explaining diagnostic substitution  
  55
- Deinstitutionalization and the variation in autism rates  
  58

2  
**The Feebleminded**  
  76

3  
**The Surveillance of Childhood**  
- The unification of mental deficiency and mental hygiene under child psychiatry  
  85
- The role of the middle-class family  
  89
- The institutionalization of children and the comprehensive surveillance system  
  91

4  
**Deinstitutionalization**  
- A new look at the deinstitutionalization of the retarded  
  98
THE AUTISM MATRIX

The middle-class family and the “valorization of retarded existence” 105

5  “An existence as close to the normal as possible”:
   Normalization 111
   Normalization as therapeutic practice 114
   Behavior modification and normalization 122

6  Childhood Schizophrenia 127

7  The Rise of the Therapies 141
   Autism therapies in the 1950s and 1960s 143
   Working on the child’s brain 148
   Inculcating habits and building skills 151
   Blurring the boundary between expert and layman 153
   The space between fields 161

8  Bernard Rimland and the Formation of NSAC 167
   The parent-activist-therapist-researcher as a new type of expert 172
   The problem of credibility 176
   Schopler and the new economy of blame and worth 178
   Autism parenting as a vocation 183
   The implications of behavioral therapy 186
   Conclusion 190

9  The Atypical Children 194
   The struggle over inclusion in the Developmental Disabilities Act 197
   Autism as concurrent with mental retardation 199
   Looping and the transformation of autism 205

10 Asperger and Neurodiversity 212
    The riddle of simultaneous discovery dissolved 213
    “Personality trait” vs. “psychotic process” 216
    Twins reunited 223
    Loops of self-advocacy 226

11 The Space of Autism Therapies and the Making and Remaking of the Spectrum 234
    The return of Rimland 236
    The agonistic network 239
CONTENTS

Blurring the boundary between medicine and alternative medicine 244
The space of autism therapies 250

Conclusion 257

Notes 270
References 287
Index 307
ACKNOWLEDGEMENTS

Many individuals provided us with useful comments after reading drafts of the manuscript or attending presentations of its main argument and findings, others have helped us through various stages of the research that led to this book. Thanks are due to: Peter Bearman, Dalia Ben-Rabi from JDC-Brookdale Institute (Israel), Douwe Draaisma, Vivian Ducat, Dani Eshet, Dahava Eyal, Ayelet Fischer, Marion Fourcade, Gyorgy Gergely, Kim Gilbert, Robin Gurley and Marguerite Kirst Colston at The Autism Society of America, Ian Hacking, Barbara Harmon, Haim Hazan, Noa Herling and Dafna Erlich from ALUT (Israel), Dagmar Herzog, Marissa King, Eric Klinenberg, Andrew Lakoff, Tanya Luhrmann, Carol Markowitz, Xue Ming, Anne Montgomery, Dan Navon, Ari Ne’eman, Molly Ola Pinney, Beatrice Renault, David Schelly, Chloe Silverman, Regina Skyer, Kathy Small, Michael Staub, Rachel Tait, Zsuzsanna Vargha, Lynn Waterhouse, Juliette de Wolfe, participants in Gil Eyal’s Sociology of Expertise seminar at Columbia University, and the many parents who graciously took the time to share their stories and opinions with us. Thanks are also due to the editorial team at Polity Press. John Thompson shepherded the whole process with a sure hand and sage advice, and Ann Klefstad read the whole text and made useful corrections designed to make the book more communicative. Part of the research for this book was conducted with the generous support of the National Science Foundation under Grant No. 0719823. Any opinions, findings, and conclusions or recommendations expressed in this book are those of the authors and do not necessarily reflect the views of the National Science Foundation. Some of the manuscript was written while Gil Eyal was resident fellow at the Center for Advanced Studies in Stanford, California. Our thanks to the Center’s director Claude Steele and deputy director Anne Petersen for their generous support.

viii
INTRODUCTION: THE AUTISM MATRIX

Autism has become highly visible. Once you begin to look for it, you see it everywhere. Search the archived pages of major newspapers, and you will find countless reports on scientific advances in the detection, understanding, and treatment of autism; letters to the editors protesting insurers’ miserly reimbursement for treatment options; dispatches from the battlefronts of special education; stories of parents’ love and determination in the face of tragedy, of their dogged advocacy for their children. Type the keyword “autism” into Yahoo! Groups and you will get 3,072 entries, from “Texas-Autism-Advocacy” to HBO (hyperbaric oxygen) therapy for autism. Many of these sites contain lively discussions among parents, professionals, and other interested parties who have banded together virtually for this purpose and are communicating across county and state lines, across national borders. Sort through your mail or take a walk in your neighborhood reading car stickers, and you will surely discover that April is International Autism Awareness Month, that not far from you there is a special preschool or after-hours program dedicated to autism. Stroll into your local library or bookstore and you will discover whole shelves dedicated to autism sourcebooks, advice to the parents of autistic children, memoirs of individuals with autism or of their parents. Then start talking to somebody who is also browsing through these titles.

A few clicks away, a simple question addressed to a stranger, and you will be introduced to a host of strange terms that, you recognize, you had heard mentioned before but preferred to ignore: most important, you will learn about “the spectrum,” about “high functioning” and “low functioning,” about “Asperger’s syndrome.” You will hear talk about GFCF diets, chelation and mercury in vaccines. You will encounter numerous references to therapies going by various
acronyms such as ABA, DIR, RDI, RPM, PRT. You will learn to refer to children as being “very sensory,” having “OT (occupational therapy) issues,” and, most alarming of all, you will begin to look at your own children, or any other children around you, differently: Do they make eye contact? How long and how well? Do they walk around on their tiptoes and like to flap their hands? Do they know how to make friends? Do they like to run their hands over the soft fabric of toys rather than play with them? Autism has not only become highly visible, it’s become the center of a social world, a universe of discourse complete with its own idioms, modes of seeing and judging, its own objects and devices. Wait but a little time, and you will begin to scrutinize not only children’s behavior, but your own, your parents and siblings, your childhood memories, for the telltale signs.

Why is autism so visible now? We all know the answer. There is an epidemic. In the U.S., the Center for Disease Control estimates that the prevalence of autism has increased from 4 per 10,000 in 1989 to 66 per 10,000 in 2002; that is, from 1 in 2,500 to about 1 in 150 children.1 A recent article in Pediatrics provided an estimate of 110 in 10,000 (1 in 91) based on parental reports (Kogan et al. 2009). Likewise, data from California show that between 1987 and 2003 the number of autism cases handled by the California Department of Developmental Services increased 634 percent. Around the globe, the incidence of autism is estimated to be rising – to 1 in 100 in the U.K., 1 in 250 in India, 1 in 1,000 in China – though the reliability of these estimates is very uneven.2 We all know what an epidemic is – a public health emergency. A serious and devastating illness is spreading rapidly in the population. We all know what needs to be done about epidemics. It’s a call for action. Detection centers must be established so new cases are rapidly identified, isolated, and treated. Money, lots of money, must be “thrown” at the scientists so they will come up with an explanation and a solution. The number of new cases must be brought down, the trend reversed.

But can this really be the answer? There are many who think it was exactly the other way around: it was not the epidemic that made autism visible, but the visibility of autism that made the epidemic. They say that changed diagnostic criteria, greater awareness, and better detection services have increased the frequency with which autism is diagnosed. We will address this argument in more detail in a few pages, but for the moment we note that from this point of view, we can think about claims of an epidemic as attempts to establish a discursive link, to throw an improvised rope bridge across from the autism social world to the worlds where decisions are made, resources
INTRODUCTION

allocated, and actions taken – the worlds of politics, economics, medicine and science. In these worlds, what do they care if children make eye contact? But they do care about epidemics; that is their business. The claim of an epidemic is salutary if it makes visible the plight of suffering children and their parents, but we think that on balance, it does more harm than good. It raises the alarm, and we must admit that we ourselves initially became interested in autism because we heard talk about an “epidemic.” We asked: why are the numbers of diagnoses rising? What are the processes and causes leading to the current autism epidemic? Only slowly and painfully did we come to realize that this was a simplistic question. The search for the epidemic’s cause was as good a place as any to start, but ultimately blinkered our vision. The claim that there is an epidemic constrains the communication between the autism world and these other worlds where decisions are made. As a discursive link it is self-defeating. It provokes a futile and barren debate about whether there is an epidemic or not (Shattuck and Durkin 2007), whether it is “socially constructed” or real. Are new cases evidence of children poisoned by vaccines or of an overzealous psychiatric profession?

In this book, we would like to change the terms of the debate. We would like to provide a different interpretation for the rise in the number of autism diagnoses and a different way of establishing a discursive link between the autism world and its interlocutors. The current rise in autism diagnoses, we argue, should be understood as an aftershock of the real earthquake, which was the deinstitutionalization of mental retardation that began in the late 1960s. The deinstitutionalization of mental retardation was a massive change, not only materially – large institutions emptied, some razed to the ground, some converted into more humane service centers – but also symbolically. Deinstitutionalization acted as a sort of “moral blender” into which disappeared the old categories that reflected the needs of custodial institutions (moron, imbecile, idiot, feebleminded, mentally deficient, mentally retarded – whether deemed educable or trainable, or neither – emotionally disturbed, psychotic, schizophrenic child, and so on). The moral blender of deinstitutionalization scrambled these categories, giving rise to a great undifferentiated mass of “atypical children” (we will explain later why we are using this term). Then, gradually, new categories began to be differentiated within a new institutional matrix that replaced custodial institutions – community treatment, special education, and early intervention programs. It is this institutional matrix and the therapies that populate it which gave rise to our current notion of a spectrum of autistic disorders running
the whole gamut from children with severe disabilities who speak little and require round-the-clock care to semi-genius teenagers with Asperger’s disorder. Beginning in Chapter 2, after we have laid the necessary groundwork of the book’s argument, we will give a historical account of this matrix and how it was assembled.

As well as giving rise to the new institutional matrix, and intimately connected to it, the deinstitutionalization of mental retardation signaled a massive change in the social organization of expertise. It was pushed forward by challenges to psychiatry from relatively marginal groups – special educators, occupational therapists, behavioral psychologists, activist social scientists identified with the anti-psychiatry movement – and parents. In the U.S., the National Association for Retarded Children (NARC) played a crucial role, and as we shall see in later chapters, so did the National Society for Autistic Children (NSAC) – both parents’ groups. They all sought to undermine the dominance of the psychiatric profession, which had in the past ignored or belittled their expertise. Rather than unseating or replacing psychiatry, the challengers maneuvered around and beneath it, so to speak. In doing so, they opened up a vast contested space between professional jurisdictions into which entered all sorts of entrepreneurs from adjacent fields – psychiatrists, no doubt, but also academic psychologists, occupational therapists, speech therapists, special educators, activists, and parents – each peddling a different typically low-tech therapy tailored to suit the needs of the new institutional matrix. In the past, psychiatrists used to rule the roost by virtue of their monopoly over the directorship of large state institutions for the mentally retarded. The new institutional matrix of community treatment, special education, and early intervention, however, acts as a great leveler, putting the psychiatrist on equal footing with occupational therapists and special educators, since all must appeal to and strike an alliance with parents. In the custodial matrix, the psychiatrist acted in loco parentis, absolved the parents of their responsibility and expropriated their knowledge.3 To succeed in the new institutional matrix, on the other hand, one must pay homage to its oft-repeated mantra that “parents are experts on their own children” and make room for meaningful interchanges between their expertise and one’s own. Psychiatrists were the least prepared to adapt to such changes. No surprise, then, that they were outmaneuvered by swifter opponents.

To substantiate these arguments, we will first analyze, in Chapter 1, the international variation in autism prevalence rates, as well as between U.S. states, and show that it systematically correlates with indices of deinstitutionalization and parent activism. The bulk of
what follows through Chapter 11 is historical analysis of how the autism spectrum became the preferred way to represent and intervene in childhood disorders, showing the contribution to this process of deinstitutionalization, parental activism, and the reorganization of expertise. The analysis in this book is based mostly on texts: articles, published interviews, reports, and manuals. We also conducted interviews with parents, therapists, and advocates, and participant observations at therapy and diagnostic centers. We will analyze this data in depth in our second book, which will focus on the moral career of the autistic child. Occasionally, however, we will use data from interviews and observations to illustrate some of the arguments in this book.

So what does the deinstitutionalization of mental retardation have to do with autism after all? One can imagine the psychiatrists being relieved by no longer having custody over mental retardation. There was never much profit or prestige in it anyway, only trouble. The mentally retarded were severely underfunded and highly stigmatized, and the stigma infected whoever was in charge of them. Mental retardation was uninteresting for psychiatrists since there was very little to do about it. It should not have been the charge of psychiatry – whose role is to treat mental illness – to begin with. Isn’t autism mental illness? Doesn’t its story begin in 1943, before the deinstitutionalization of mental retardation, when the leading child psychiatrist Leo Kanner discovered it? Shouldn’t we trace autism’s origin back to the moment Kanner wrote his landmark article “Autistic Disturbances of Affective Contact” and introduced the world to a curious group of eleven children he had seen in his Baltimore practice, the first eleven people diagnosed as autistic in history? Unlike mental retardation, autism was always exciting for the psychiatrists, a mysterious disorder through which they hoped to catch a glimpse of how the normal mind/brain works (Rimland 1964, 3). They fumbled around for a while, no doubt, misled by psychoanalytic dogmatism and charlatanism. But as psychiatry became more scientific, more securely founded in neurochemistry, genetics, and brain imaging, it took up again Kanner’s sound empirical observations, followed the thread untangled by his clinical wisdom, and is now hard on the heels of an explanation. Soon it will identify the genes and brain mechanisms involved and will devise a treatment. In the meantime, it has reiteratively modified diagnostic criteria to reflect its better understanding of the disorder, thereby producing the rise in the number of diagnoses.

One notes, of course, that it is not at all clear or self-evident why a better scientific understanding of autism should produce a dramatic rise
Digression: On terminology

Why are we using this offensive term “mental retardation”? Hasn’t it been replaced by a more sensitive vocabulary? Throughout this book we refer to “mental retardation,” “mentally retarded” children, “the mentally retarded” and, worse still, to “retardates,” “morons,” “imbeciles,” and “idiots” (we further explain the latter three terms in Chapter 9). Where it is historically accurate, we also employ the term “intellectual disability,” as well as other terminology now in fashion. Words like “mentally retarded” may be unpalatable to many readers. These labels might have been used to deprive a loved one of services, or to devalue the richness of his or her experience. Our use of them is purely historical. In other words, we use the terms as they were used by researchers and physicians at particular historical moments. There are sharp differences in the moral images and ethical responses associated with these different terms and the sense conveyed by each has shifted dramatically over time. This book considers how these categories – autism and mental retardation in particular – have rearranged in relation to one another over the span of little more than half a century.

Let us be clear here. We are not claiming that people who are really mentally retarded have secretly infiltrated the autism spectrum. Nor are we suggesting that people who were truly autistic were mislabeled as mentally retarded in the past. We are arguing, rather, that the ways in which we think about and deal with childhood and developmental disorders, the very distinctions that we make, are what have changed. In the wake of deinstitutionalization, mental retardation sat at the center of the emergent domain of “developmental disabilities” (see Chapter 9) as the prototypical form of developmental disability. We argue, however, that it has lost this centrality in favor of autism, which is gradually coming to occupy the bulk of this domain as a multidimensional spectrum of pervasive developmental disorders. Mental retardation qua intellectual disability is now on the margins of this domain.

Today we grimace at the term “mental retardation,” and prefer euphemisms such as “intellectual disability” or “developmental disability.” Yet, when the term “mental retardation” was first used it was itself introduced as a euphemism. It was originally invented to denote those with only a mild deficit, those who were merely retarded rather than “idiots” or “imbeciles.” In fact, it took over from terms such as “mental deficiency” or “feeblemindedness,”
each of which in their turn went through the same cycle. Each began as a euphemistic term for those slightly below “normal,” then extended in a benevolent gesture to cover the whole realm of “mental defect,” by virtue of which each gradually acquired a pejorative and ugly connotation which made it less and less useful. Then a new euphemism would be invented and the whole cycle would begin again (Wallin 1949, 5–6).

Before ending this digression, we must mention one other point of contention. Some readers may feel we should use “person-first” language. We refer at times to “autistic children” and even “autistics.” Some disability studies scholars and advocates have argued that we ought to put the person first, before the label. We should refer to “people with disabilities” or more specifically in our case “a child (or adult) with autism.” However, in deviating from person-first language at certain points, we are following autism advocates and accentuating a point about the novelty of autism. Parent-activist-researchers, like Bernard Rimland (1993), have argued against person-first language because they think “autistic child” conveys how much autism pervades every aspect of their child’s experience better than “child with autism.” Jim Sinclair, a leading self-advocate, has also eloquently described why he himself does not use person-first language. “Saying ‘person with autism,’” he writes, “suggests that the autism can be separated from the person. But this is not the case . . . If I did not have an autistic brain, the person that I am would not exist. I am autistic because autism is an essential feature of me as a person” (1999). Finally, we follow this use of terminology because it expresses a key aspect of the autism discourse we are describing: autism, on this view, is not something added to a person, like a disease that you can catch and later cure; it is a way of being that pervades every aspect of one’s experience in the world.

in the number of diagnoses unless one assumes that the condition has been misdiagnosed in the past and was hiding under some other designation. This leads one back to mental retardation and to the mounting evidence – discussed in Chapter 1 – that a great deal of the rise in the number of autism diagnoses was caused by diagnostic substitution from mental retardation. So we are back to square one, to the deinstitutionalization of mental retardation. It created the institutional conditions of possibility for this diagnostic substitution, the ground upon which our current mode of representing and intervening in autism took
shape. We have grave doubts whether psychiatry or a better scientific understanding had very much to do with this transformation.

We became interested in the deinstitutionalization of mental retardation because we noted two simple facts about autism. First, since there is still no medical treatment for autism, the bulk of the work in this field is done by paraprofessionals deploying behavioral, speech, occupational, or other therapies. Psychiatrists diagnose autism and (increasingly) prescribe medications, but these are merely meant to control mood or undesired behaviors, not to treat the core of the disorder. The bulk of therapeutic work – even though some of it is characterized as “biomedical” – escapes the jurisdiction of psychiatrists, though they may endorse it or even undertake to provide it themselves.

Second, as we shall show in Chapter 7, many of these therapies were not originally developed for treating autism but for mental retardation. More precisely, as we came to recognize once we delved into the history of these therapies, what they have in common is that they blur or hybridize the boundary between mental retardation and mental illness. Some, like Sensory Integration Therapy (SIT), originated in the field of mental retardation yet projected themselves outside it because they treated retardation as “brain injury,” a semi-illness that could be cured. Others, like Applied Behavior Analysis (ABA), worked in the opposite direction: originating in the field of mental illness, they projected themselves outward because they treated illness as a bad habit or a lack of skills that could be corrected by means of behavior modification. These therapies date from the mid-1960s when they were fairly marginal. Certainly psychiatrists had very little to do with their invention or diffusion. Their moment came with deinstitutionalization. Their blurring of the boundary between mental illness and retardation corresponded to the space that deinstitutionalization opened up between professional jurisdictions. They became an integral part of the new institutional matrix of community treatment, special education, and early intervention.

So we had a hunch: what if “idiocy,” “feeblemindedness,” “mental retardation,” “autism,” “the spectrum,” are all interpretations we superimpose over the longue durée of practices, and the real events take place not with the proliferation of this or that interpretation (such as the autism epidemic), but when a whole institutional matrix of practices gives way to another? This is precisely why we say that the question about the causes of the epidemic is simplistic, since it ignores this more subterranean change. The issue is not whether the rise in the number of diagnoses is due to vaccinations, pollution, or diagnostic substitution, whether it is “real” or fabricated. The issue is that our
practices for representing and intervening in childhood disorders are no longer constrained by the opposition between retardation and illness, but proceed as if they can ignore it. What is the significance of this change? This is where the discursive link, the bridge between the autism world and its interlocutors, should be built.

So powerful, however, are the terms in which the current debate is joined, that we have no choice but to address them first, before we attempt to explore the significance of this blurred boundary between retardation and illness. Are we saying that there is no autism? That there is no epidemic? That autism is not real? That it’s all “socially constructed”? Are we denying that autism is a real disorder, rooted in the neurochemistry of the brain? Or worse still, are we merely splitting philosophical hairs with no relevance to the actual suffering of children and their parents? The following section attempts to clarify the approach we are taking in this book and to indicate how it relates to existing explanations for the autism epidemic – why it is legitimate, we think, to emphasize the role played by therapies and institutional factors, and why it does not involve a denial of the reality of autism. Only once we have dealt with this issue and demonstrated that the terms of the current debate are imprecise and unproductive can we return in the concluding section of the introduction to drawing out the implications of our argument: what do we learn about autism and its history by connecting it with the deinstitutionalization of mental retardation?

**Why focus on therapies?**

To summarize our argument this far: the rise in the number of autism diagnoses is an indirect product of deinstitutionalization. The new institutional matrix of community treatment, special education, and early intervention that developed in its wake, and especially the very therapies that were meant to treat autism within this matrix, gave rise to a spectrum of autistic-type disorders that straddles an indeterminate terrain between mental illness and retardation, thereby laying the groundwork for the epidemic. The reader, quite justifiably, may suspect that this claim is another in a long series of “debunking” arguments made by social scientists, of which Szasz’s *The Myth of Mental Illness* is perhaps the most notorious. Our contract with the reader, upon entering this section, is that we show that the focus on therapies is not a denial of the reality of autism. We would like to show, in fact, that the sociological approach we have crafted, with its focus on therapies, is better suited to accept without quotation
marks the specific reality of autism and the autism epidemic, whereas the nonsociological approaches of medical researchers, what we call “naturalist explanations,” inevitably lead to a controversy over the reality of autism and the autism epidemic.

By “naturalist” we mean arguments of the following type: “The rise in the number of autism diagnoses is an accurate reflection of an actual rise in the number of real autism cases out there, caused by . . . .” There are different versions of this type of argument, and readers are invited to pick their poison. Some researchers have been looking for evidence of correlation between rising autism prevalence and environmental toxicity (Roberts et al. 2007). A group of parents has created a great deal of debate and controversy by linking new autism cases with a mercury-based preservative in vaccines (Bernard 2004). Other researchers have linked autism to TV watching (Waldman, Nicholson, and Adilov 2006). All of these explanations work on a “pollution” model, which some anthropologists characterize as an ancient moral narrative of accusation and blame (Douglas 1966, 1992). Conversely, other researchers and commentators assure us that the epidemic is “nobody’s fault,” by attributing the rising numbers to a genetic inheritance increasingly favorably selected in an age dominated by communications and information technology (Baron-Cohen 2006). It should be emphasized, however, that the bulk of naturalist research on autism does not touch at all on the question of rising prevalence, seeking merely to establish connections among autism, genes, and brain mechanisms. Those biologists and geneticists are content to endorse a social constructionist explanation for the epidemic, though vaguely and noncommittally insinuating that some combination of multiple genes in interaction with environmental factors or pollutants could have brought about some increase in prevalence.

No matter who or what they blame, or whether they avoid accusation altogether, all naturalist explanations face the same two problems, which cast doubt on their plausibility as explanations for rising prevalence:

First, the actual population of autism diagnoses is extremely heterogeneous. Until now we have been using the term “autism.” We have neither been quite accurate, nor quite up to date. The correct term is “autism spectrum disorders.” The DSM-IV classifies autism, or more precisely autistic disorder, as a “pervasive developmental disorder” together with Rett’s disorder, childhood disintegrative disorder, Asperger’s disorder, and pervasive developmental disorder—not otherwise specified (PDD-NOS). The latest word from the National Institute of Mental Health is that “all children with autism spectrum disorders” . . .
disorders demonstrate deficits in 1) social interaction, 2) verbal and nonverbal communication, and 3) repetitive behaviors or interests.” That’s quite broad. Admittedly, the numbers reported above were counts of autistic disorder by itself, without the other four conditions, but one should note, first, that the prevalence of Asperger’s disorder and PDD-NOS has been rising in tandem with autistic disorder, and second, that the boundaries between autistic disorder, PDD-NOS, and Asperger’s disorder are ill-defined and may change in accordance with pragmatic considerations.

Even if we look only at autistic disorder and exclude the other four, the heterogeneity remains enormous, because the diagnostic criteria are now organized in a way that reflects the idea of a “spectrum.” The reader may consult Figure 1 below, which reproduces the DSM-IV-TR diagnostic criteria for autistic disorder. A child who meets six out of twelve criteria – that is, a child who is quite possibly verbal, affectionate, and friendly, but who does not engage in typical social play, and whose range of interests is rigid (1bc, 2bd, 3bd), would be diagnosed as autistic. The same holds, of course, for a child who meets all twelve criteria, struggles to communicate at all, and seems unaware of the presence of others. As a result, the spectrum includes children who do not speak and are profoundly disabled, together with semi-genius teenagers and the whole range of in-between cases. The spectrum includes many children who are concurrently diagnosed with mental retardation, but also many whose IQ scores are well above normal. Nobody really knows the true extent of co-morbidity with mental retardation. Estimates vary widely, from 31 to 36 percent in a California study (Croen et al. 2002a; 2002b) to upwards of 89 percent reported by Dutch and Swedish researchers (Kraijer 1997, 40–42). Calling these “estimates” is misleading. They are more like prescriptive statements than descriptive ones. They reflect, in fact, a disagreement about the very nature of the disorder, what should count as autism, and what is to be understood by the metaphor of a “spectrum.” The same heterogeneity is true for other co-morbid conditions as well. Many children with autistic disorder are also diagnosed with epilepsy, hyperactivity, emotional disorder, learning disability, or some combination thereof, but many are not.

As we shall see, a sociological explanation can coexist quite comfortably with such diversity. But the scientists, the medical researchers advancing naturalist explanations, cannot. If you conscientiously seek a naturalist explanation for autism and the autism epidemic, you must be deeply troubled by such mess, and indeed, many geneticists and brain researchers are deeply troubled. In their view, autism is not “a
single syndrome with highly variable severity (the autistic spectrum),” but “an aggregate of specific disorders that share some common [behavioral] features” because they involve the “dysfunction of one or more, probably widely distributed, brain systems” (Waterhouse et al. 2007, 308). To figure out “the patho-physiology of autism” – which brain systems are involved and through what causal pathways they are affected – “requires the development of a validated typology based on
behavioral criteria.” But, protest the scientists, there is no consensus on such a typology, and the spectrum is defined far too broadly to do the job (Rapin 1994). Consequently, the “inclusiveness” of the spectrum “has become a critical problem for genetic and brain research in autism” (Waterhouse et al. 2007, 308). Simply put, when they pick subjects for an autistic experimental group and a non-autistic control group, to determine if they differ in terms of genes, fMRI profiles, or what have you, they are never sure that experimental subjects in fact share the same condition. Now, our point is not to say that this makes medical research into autism necessarily impossible. This semi-vicious circle we describe is not unique to autism, but common to many psychiatric diagnoses, because they typically lack an objective biological marker (like trisomy-21 used to identify Down’s syndrome), and yet there is no denying that research has produced great advances in the understanding and control of such conditions.7 Our point, however, is to indicate that the logic guiding this research, the logic of naturalist explanation, of necessity pushes it in the direction of deeply distrusting the reality of the autistic spectrum and dissolving its unity. In the absence of a validated behavioral typology or a biological marker, and since any etiological typology “does not encompass the great majority of children in whom etiology remains unknown” (Rapin 1994), researchers of necessity “slice” the spectrum this way or that in order to get meaningful correlations with genes, brain structures, pollutants, and so on. As Harvard geneticist Christopher Walsh puts it: “I would like every child on the spectrum to have not ‘autism,’ but a more specific disorder” (Pettus 2008). This means that it is they, the scientists, not us, the sociologists, who must argue that some kinds of autism are not “really” autism, or that autism, as we heard above, is not a “single syndrome” but an “aggregate of specific disorders,” which really means that they think that autism or the autistic spectrum is merely a provisional label, soon to be replaced by more specific and better specified knowledge.8 For example, the New York Times reports that “with technology that can now scan each of an individual’s forty-six chromosomes for minute aberrations, doctors are providing thousands of children lumped together as ‘autistic’ or ‘developmentally delayed’ with distinct genetic diagnoses.” The article continued to report on six children, all with autistic symptoms, who have been knit together into a separate condition named “16p11.2” after the chromosomal address of their defective DNA (Harmon 2007). The quotation marks in the piece say it all. It is not some sociological mumbo-jumbo, but the very logic of scientific inquiry that is forcing a controversy over whether autism is really real or just a provisional label, whether the epidemic is real or socially constructed.
Is the argument of this book contradicted by the well-documented evidence concerning a genetic component in autism? Below we try to unpack the idea of autism as a genetic disorder and ask what it means. We try to show that while little is known at the moment about even the most basic questions, the thrust of genetic research is, if anything, supportive of an explanation of the rise in autism diagnoses due to a complex historical transformation that involved diagnostic expansion and substitution with mental retardation.

**How much of autism is genetically inheritable?**

Folstein and Rutter’s 1977 monozygotic twins study established that autism is genetically inheritable, and led to subsequent studies that estimated the genetic heritability of autism to be very high, around 90 percent (Bailey et al. 1995). A heritability estimate is a population measure of the proportion of the overall phenotypic variance attributable to genotypic variance (typically by contrasting risks of monozygotic and dizygotic twin when the other twin has the phenotype). The problem is that all these studies were based on fairly small samples (21 in the original 1977 study) typically obtained through referrals, which made their estimates unreliable and biased upwards. A new study drawing on a much larger random population sample estimates heritability at 19 percent for males and 63 percent for females (Liu, Zerubavel and Bearman 2009). Ultimately, therefore, how much of autism is genetically heritable is simply unknown. It is possible, moreover, that there is no single number to be had, that the genetic heritability of autism is a moving target. In the time that elapsed between 1977 and now, as we show in this book, the autism spectrum has expanded, thereby likely making the autistic phenotype less specific and decreasing the genetic heritability of the condition.

**Does genetic research resolve Waterhouse’s question of whether autism is a single syndrome or an aggregate of specific disorders?**

The short answer is no. The longer one is that on balance genetic research has come up with a great deal of evidence contrary to the idea that autism is a single syndrome of varying severity and instead has indicated that at the genomic level the boundary between autism and MR is blurred. What is this evidence?
Researchers trying to assess the genetic heritability of the three core deficits in autism spectrum disorders (social, communication, and repetitive behaviors / restricted interests), using a twin study design, found only modest co-variation between them. Individuals with extreme scores in one domain did not have extreme scores in others. Consequently, they now study each domain separately, finding each is traceable to a different quantitative trait locus (QTL) with little overlap between domains or studies, underscoring “the importance of genetic and phenotypic heterogeneity in ASDs [autism spectrum disorders]” (Abrahams and Geschwind 2008, 342; Ronald et al. 2006; Alarcon et al. 2002; Schellenberg et al. 2006; Chen et al. 2006; Szatmari et al. 2007).

The search for QTLs is intimately connected with the attempt to specify endophenotypes that would put genetic research on autism on a more sound footing. Endophenotypes are a sort of intermediate phenotype. They underlie more overt symptoms; they are present in an individual whether or not the illness is expressed; they are found in non-affected family members at a higher rate than in the general population; and they have a clear genetic component. Up till now, the most common endophenotype found to be associated with autism is language delay, quantified as age at first word and associated with variation in the CNTNAP2 gene. Language delay, however, is an endophenotype that obviously is not unique to autism but common to “other clinically distinct, but related, disorders,” including intellectual disabilities, specific language disorder, and other conditions, thereby rendering autism less of a single well-bounded syndrome and leading researchers to suggest that “current clinical notions of boundaries between neuropsychiatric disorders need not be representative of the underlying genetic or biological etiologies” (Abrahams and Geschwind 2008, 350, 352–53; Alarcon et al. 2002; Bolton et al. 1994).

The genetic and phenotypic heterogeneity of autism spectrum disorders – which Abrahams and Geschwind (2008, 350) explain is due in part to aggregating samples from various groups throughout the world, as well as to differences in diagnostic criteria across both space and time has led other geneticists to a slightly different research strategy that reduces this heterogeneity by subsetting the spectrum into smaller, more distinct phenotypes, for which a more specific genomic linkage may be sought. By thus splitting the spectrum into more cohesive clusters of children who, for example, experienced normal development before onset of autistic regression, or who suffer also from seizures, or who exhibit extreme rigid and repetitive behaviors, researchers
were able to identify new loci on the genome associated with a cluster, but not with other autism spectrum cases (Abrahams and Geschwind 2008, 350; Cantor et al. 2005; Stone et al. 2004; Molloy et al. 2005; Buxbaum et al. 2001; 204; Shao et al. 2002).

A different though potentially complementary research strategy has been to look for rare mutations that are common to large groups of individuals on the spectrum. This approach has been productive – perhaps too productive. One authoritative survey found that “defined mutations, genetic syndromes and de novo CNV\(^\text{11}\) account for about 10–20% of ASD cases.” Yet, the authors hasten to add that “the striking finding” is “that none of these known causes accounts for more than 1–2% of cases,” and that none of them “consistently results in autistic disorder, Asperger syndrome or any other defined spectrum disorders” (Abrahams and Geschwind 2008, 341, 350). In other words, any one of these mutations appears in only a very small proportion of the spectrum, yet they may also appear in the normal population, or in individuals with other diagnoses the most important of which is, unsurprisingly, mental retardation. Consequently, the authors say that there is an “absence of clarity surrounding the specifics of the relationship between the ASDs, MR and other neuropsychiatric conditions . . . such as specific language impairment . . . [or] attention deficit hyperactivity disorder” (352–53). What we know up till now about the genetics of autism, they say, paints a picture not unlike mental retardation, in the sense that the spectrum designates a whole domain of loosely related yet heterogeneous conditions “for which there is no single major genetic cause, but rather many relatively rare mutations” (341).

Indeed, at the genome level autism and intellectual disability are linked and overlapping, something that provides indirect support for the argument of this book. Recent studies demonstrate that “some types of autistic disorder and mental retardation may have common genetic origins” (Laummonier et al. 2004; Marshall et al. 2008). Two of the best known of these types are Rett’s disorder and Fragile X. Both are very rare. Rett’s prevalence is 1 to 10,000–15,000 of female births, Fragile X occurs in approximately 1 of 4,000 males and 1 in 8,000 females. Numerically speaking, these conditions are too rare to make a significant dent in the spectrum or the epidemic, but they demonstrate how uncertain and provisional are the distinctions we currently employ. Both Rett’s and Fragile X cause intellectual disabilities as well as a range of autistic-like symptoms and both were often classified as either types or causes of autism. The genetic mechanisms of both conditions were recently identified – inherited
in Fragile X, sporadic or *de novo* for Rett’s – and consequently individuals who were in the past diagnosed as autistic may now, per Christopher Walsh’s wish, have the single diagnosis of Rett’s or Fragile X. Additionally, many other mutations or regions implicated in autism spectrum disorders are also implicated in mental retardation (Abrahams and Geschwind 2008, 352–53).

*Could change at the genetic level have caused the rise in autism diagnoses?*

The Web is teeming with attempts to provide a genetic explanation for the epidemic, but this is speculative territory where few geneticists dare (or care) to tread. The main sources for these speculations are the early *Wired* article that claimed there was an autism pocket in Silicon Valley (Silberman 2001) and Baron-Cohen’s (2006) theory of “ assortative mating” according to which autism risk is elevated by having two parents who are, in his terms, “high systematizers.” The problem with any such theory, in which the mechanism of change is inheritance of a trait or a mutation, is that it works only for long secular trends (Mingroni 2004). It is simply ill-equipped to explain large short-term spikes like the huge rise in autism rates from the early 1990s to roughly 2004. Additionally, if Liu et al. (2009) are correct and the heritability of autism is lower than previously estimated, such explanations would only capture a small portion of the increase. *De novo* mutations and deletions, on the other hand, could potentially provide a better mechanism to explain short-term change, but they of course must be combined with other mechanisms: either environmental pollution or demographic change. For example, Liu et al. (2009) demonstrate that a small but significant proportion of the rising autism caseload is caused by an increase in *de novo* genetic deletions due to rising parental age and the increased use of assisted reproduction technologies by older parents. The case for environmental factors, by comparison, is more speculative, though it has a lot of adherents. Supporters point to the large unexplained variance left over after estimating the effects of changes in diagnostic practices and argue that it must be due to environmental factors (Cone 2009). Both demographic and environmental accounts, however, suffer from the same problem. They identify mechanisms that change the actual genetic makeup of the population, but which would lead one to expect not a steep rise in autism per se, but a steady upward trend in developmental disorders across the board. Why would rising parental age or environmental pollution not increase also the rate of
mental retardation? And yet, as we shall see in the following chapter, there is evidence that as autism rates were rising, the rate of MR diagnoses was decreasing. The explanation we develop in this book (which points to diagnostic substitution as a proximate cause of the rise in the number of autism diagnoses), by comparison, is more parsimonious and targeted, and it does not require one to deny the incontrovertible evidence of the role of genetic mutations, whether inherited or de novo. On the contrary, it makes use of the evidence cited earlier about the genomic overlap between autism and MR (and possibly other developmental disorders). We do not need two or three mechanisms – one that explains change in the actual genetic makeup of the population, another that explains why this change is expressed as autism and not as other developmental disorders, and yet another that explains how and when these changes were recognized and diagnosed as autism – but only one: as diagnostic criteria change and as diagnostic substitution from mental retardation to autism takes place, these factors increase the genetic pool from which autism cases are drawn. The proportion of autism cases attributable to various (and unrelated) rare mutations, as well as the proportion of de novo mutations relative to genetic heritability, thus also increase. Stated in terms of genetics, diagnostic substitution allows us to make sense of how a population that must be relatively genetically stable could have experienced an “epidemic” of a genetic disease. It allows us to see that autism encompasses more of the population’s genetic variance than it once did, not because the population’s genetic makeup changed over a generation, but because the category of autism changed. It became a spectrum that captured a greater proportion of genetic endowments. Thus, the more of a realist one is about the impact of genes, the more one is obliged to adopt a sociological explanation for the rise in autism diagnoses.

**Diagnostic change**

Let us now turn to the second problem facing naturalist explanations: not only are diagnostic criteria overly broad, but over the last thirty years they have been continuously modified to reflect an increasingly broad understanding of autism. To begin with, in the DSM-II, autism was included within the diagnosis of “child schizophrenia,” which almost by definition made it a rare condition. In 1980, the diagnostic criteria were considerably relaxed by the publication of DSM-III, which separated autism from the diagnosis of childhood
schizophrenia and included it within the newly created category of Pervasive Developmental Disorders. This meant that the diagnosis of autism became independent of considerations of etiology, deep interpretation of symptoms, presence of delusions and prognosis, and was based strictly on the observation of “surface” behavior. But the diagnostic criteria were relaxed even more in 1987, with the publication of the revised edition of the DSM-III (DSM-III-R). The early onset (before thirty months) requirement was dropped. The diagnostic criterion of complete lack of social responsiveness was changed to simply abnormal social responsiveness. More important, the internal structure of the three main diagnostic criteria was changed in accordance with the idea of a spectrum. The symptoms in each of the three domains were arranged in decreasing order of severity, such that lack of social play, adequate speech but incapacity to engage in sustained conversations with others, plus a restricted range of interests, were enough to diagnose a child as autistic. The exponential rise in the number of autism diagnoses followed hard on the heels of these changes. If you are a firm believer in naturalist explanations, you can still try to claim that these changes in diagnostic criteria reflected a better understanding of autism, but you would be hard pressed to tell your skeptical listeners what might refute this assertion of belief, since the only way to diagnose autism is using the very same behavioral criteria that have changed, and there is no “objective” marker (no known brain lesion, no single gene mutation) to use in order to check their validity.

It is for this reason that many medical researchers have suddenly become sociologists and espoused a “social constructionist” explanation for the epidemic. Over and over again one finds them arguing that the rise in autism diagnoses does not reflect a real, natural rise in the number of cases out there, but is an artifact produced by the confluence of supply and demand forces (Prior 2003; Yeargin-Allsopp et al. 2003; Fombonne 1999; Croen et al. 2002a). On the supply side there was this relaxation of diagnostic criteria, and on the demand side, a heightened awareness of autism among parents, teachers and experts, which therefore led to increased detection (once people no longer think of autism as a rare condition – as it was under the guise of “childhood schizophrenia” – they begin to see it where they did not do so before), and a greater availability of services from 1991 onwards when autism was added to the Individuals with Disabilities in Education Act (IDEA) (once there are concrete benefits to getting the diagnosis, parents demand it and clinicians are happy to make it).
Does that mean that autism is not “real”? As Ian Hacking (1999) says, the question is meaningless unless we specify a real “what.” A real “disease entity”? Probably not, but not even the DSM assumes that it is. Real behaviors and brain processes that are nonetheless dependent on social experiences, habits and institutional processes? Why not? Thus, we are brought back to the statement made by the scientists: autism is not a single syndrome, they say, but a collection of specific disorders that share some behavioral manifestations because similar brain systems are affected. What do they mean by that? A term that has been used with respect to schizophrenia might be useful to clarify the point: “final common pathway” for distinctly different pathological processes, something like a “funnel” into which spill different chains of cause and effect (Jablensky 1999). The causes are antecedent and independent, as we were taught in Scientific Method 101, and so the causal chains lead one back to genes, prenatal trauma, infection, and pollution. But it would be meaningless to ask which is more “real” – the uncaused, primeval causes, or the final common pathway? Certainly the sociologist has no prejudice in this respect. One can ask which is more important, which is the weakest link in the chain, which provides the most efficient target for intervention. Yet it is very clear that for the scientists the first, and random, terms in the causal chain somehow possess more reality, or more accurately, that the final common pathway, the behavioral syndrome, possesses the least reality. Why? Because scientists think the former are “natural kinds” and that they can be isolated and studied as such, while the latter hopelessly mixes together the natural and the artificial, since it brings into play, at the very least, the judgment of clinicians, conventions of behavior (what we think we know about how “normal” children behave), childrearing practices, retrospective parental reports, and the agency of children.

Does that mean that the epidemic is not “real”? Here the sociologist is on even firmer ground. Unlike the geneticists and brain researchers, who in order to possess a proper object of study are forced to enter a dispute about which part of the epidemic is “really” autism, we can take as our object the total population of actual autism diagnoses, treating it as a real phenomenon in need of explanation. Unlike the scientists who are forced to exclude the changes in diagnostic criteria, the increased awareness, the inclusion in legislation, or the increased availability of services, as “artificial” factors, not really germane to the question at hand, we need make no such distinctions as we seek the actual processes that produce the actual population of autism diagnoses. Unlike the medical researchers, who are therefore forced,