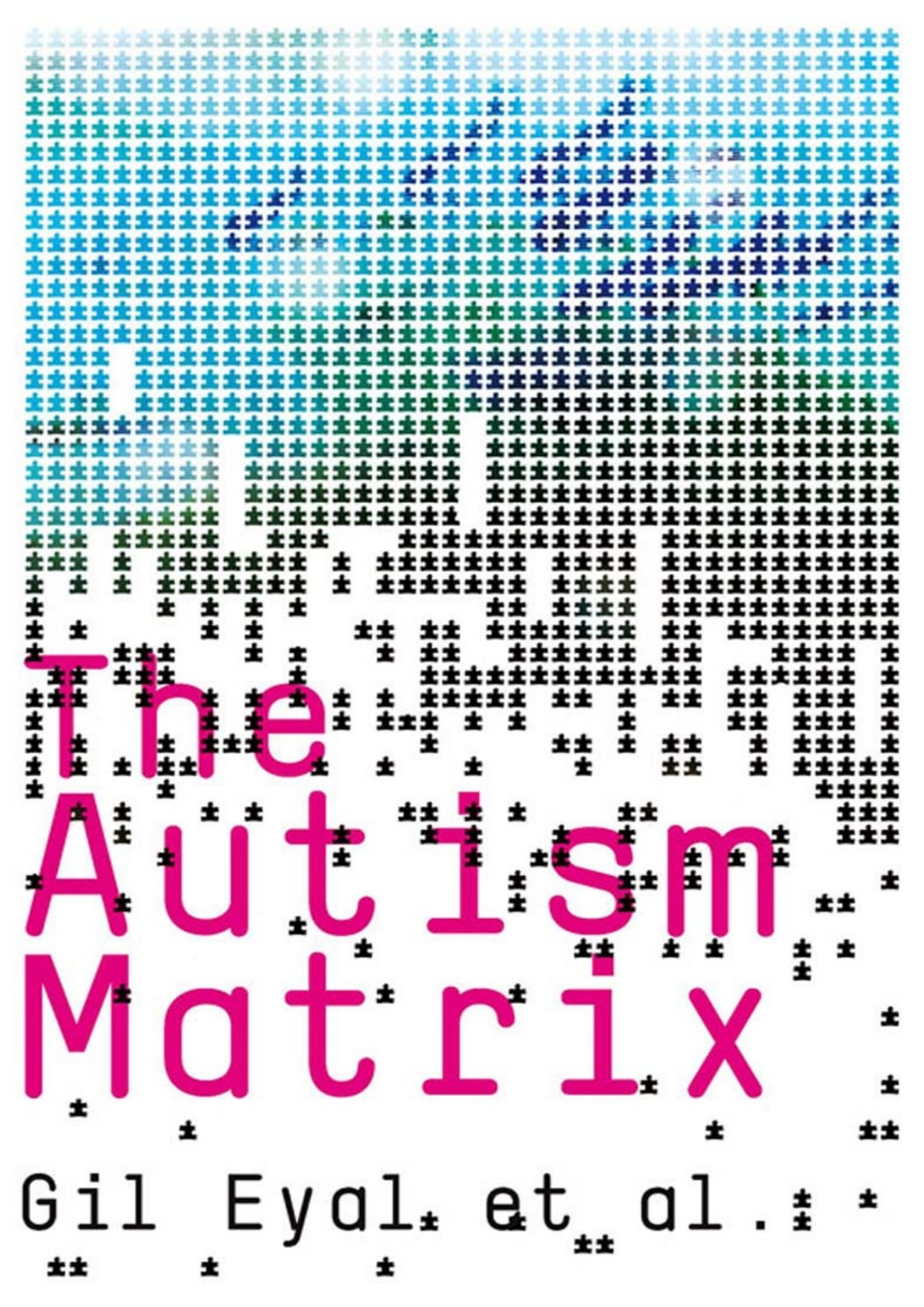
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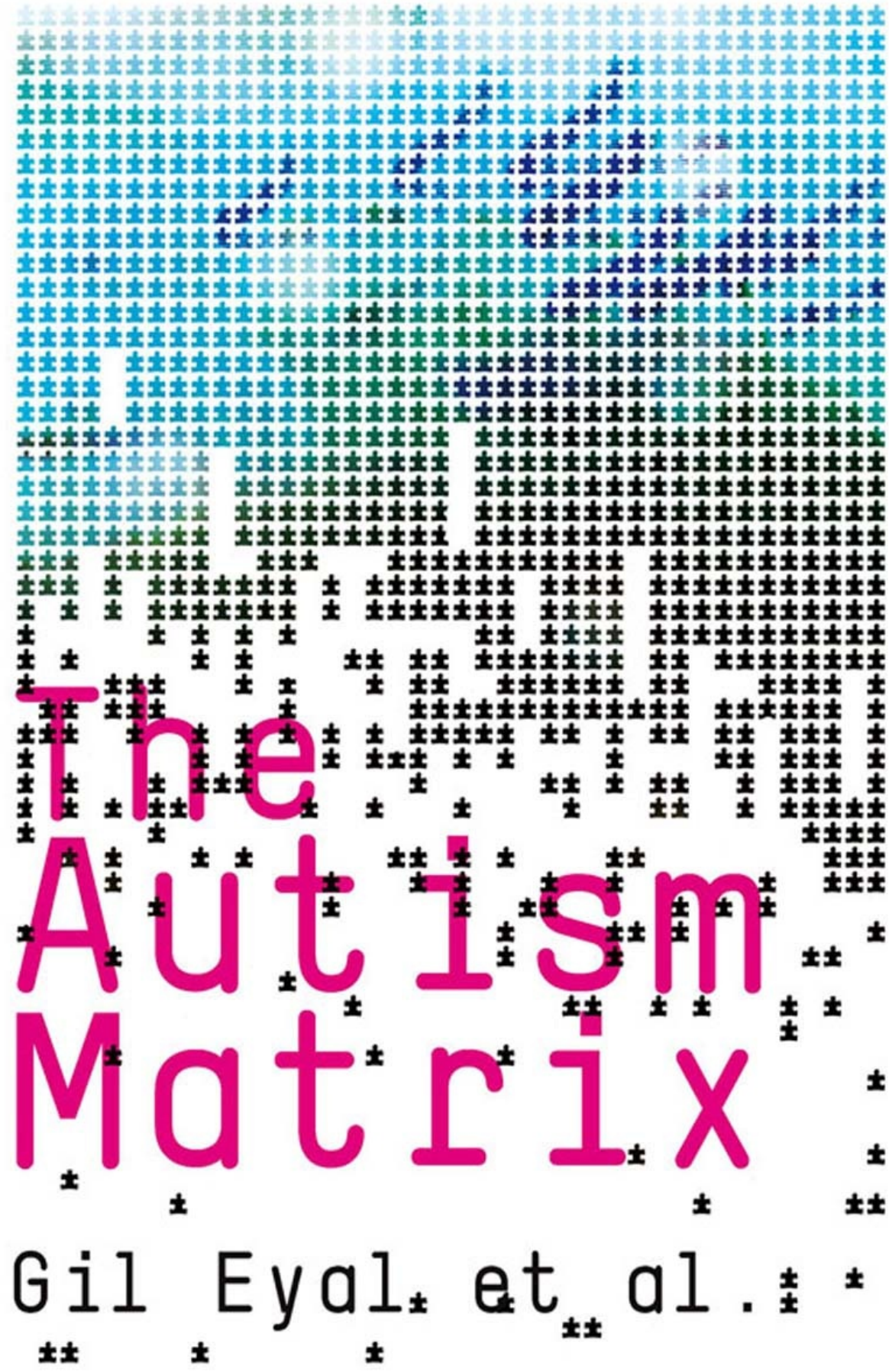
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The Social Origins of the Autism Epidemic

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polity

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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 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 ointnrepeated 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 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.

Digression: On terminology

Why are we using this offensive term "mental retardation"? Hasn't it been replaced by a more sensitive vocabulary?[4] 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.[5] 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.

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:

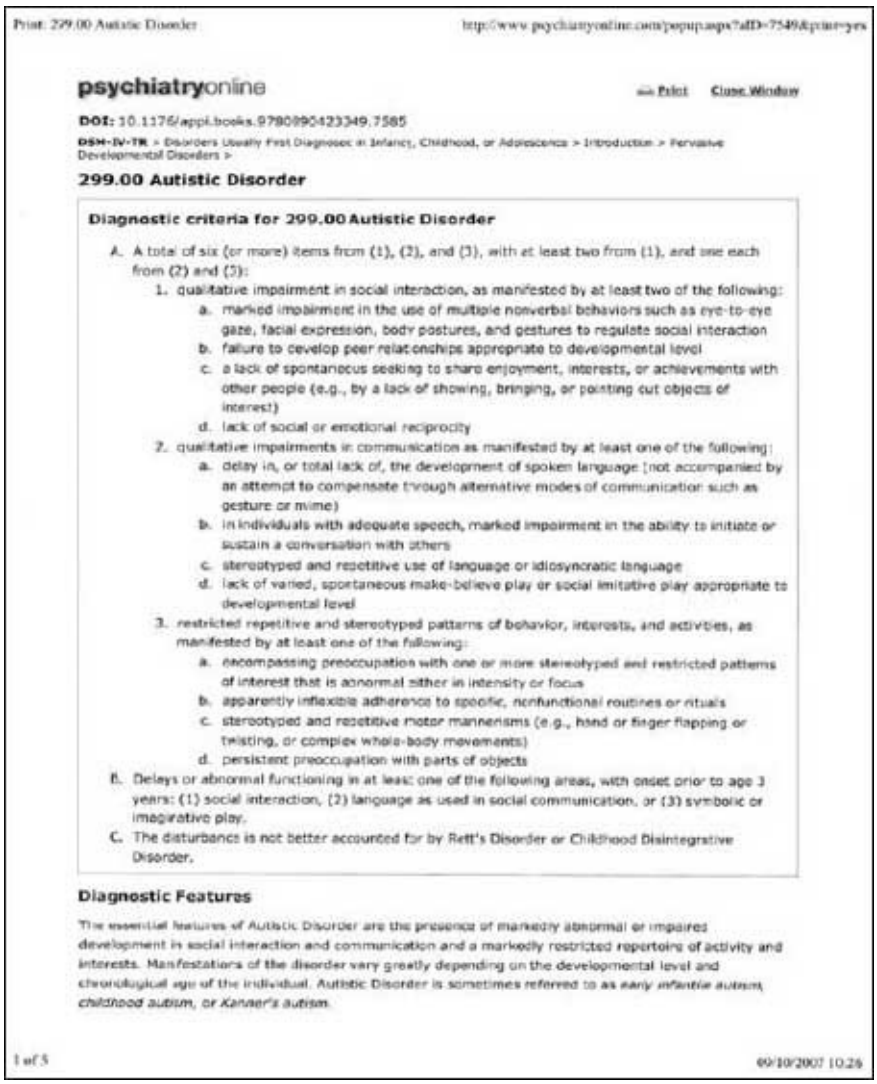
First, the actual population of autism diagnoses is extremely heterogeneous. Until now we have been using the term “autism.” We have neither been quite accurate, nor quite up to date. The correct term is “autism *spectrum* disorders.” The DSM-IV classifies autism, or more precisely autistic disorder, 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 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-IVTR 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.[6] 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.



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DSM-IV-TR > Disorders Usually First Diagnosed in Infancy, Childhood, or Adolescence > Introduction > Pervasive Developmental Disorders >

299.00 Autistic Disorder

Diagnostic criteria for 299.00 Autistic Disorder

A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):
   1. qualitative impairment in social interaction, as manifested by at least two of the following:
      a. marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
      b. failure to develop peer relationships appropriate to developmental level
      c. a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
      d. lack of social or emotional reciprocity
   2. qualitative impairments in communication as manifested by at least one of the following:
      a. delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
      b. in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
      c. stereotyped and repetitive use of language or idiosyncratic language
      d. lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
   3. restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
      a. encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
      b. apparently inflexible adherence to specific, nonfunctional routines or rituals
      c. stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
      d. persistent preoccupation with parts of objects

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play.

C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

Diagnostic Features

The essential features of Autistic Disorder are the presence of markedly abnormal or impaired development in social interaction and communication and a markedly restricted repertoire of activity and interests. Manifestations of the disorder vary greatly depending on the developmental level and chronological age of the individual. Autistic Disorder is sometimes referred to as *early infantile autism, childhood autism, or Kanner's autism*.

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Figure 1: DSM-IV-TR Diagnotic Criteria for Autistic Disorder

The genetics of autism

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)[9] 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[10] 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[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