A couple of years ago, I was asked about autism by a journalist from the New York Times. The reporter had been travelling the world, discussing the subject with many people who had contributed to our understanding of this fascinating and complex disorder. Why, she asked, was it that this subject aroused such passions? Never before had she come across a group of scientists who criticized each other quite so much as those working in the field of autism. She was astonished, dismayed and perplexed in equal measures.

This controversy forms the backdrop to a recent review of the history of autism, written by Adam Feinstein (2010). His story begins in the birthplace of so many ideas in psychiatry, Vienna in the 1930s. Here, in 1938, Hans Asperger gave a lecture on autistic psychopathy some years before the landmark paper by Leo Kanner (1943), which is often credited with initiating the avalanche of research that followed. Kanner was also born in Austria, but his parents emigrated to Germany when he was still a child, and he left for the USA as a young doctor in the 1920s.

The origins of the term autism are therefore mired in controversy, with some claiming that Kanner plagiarized Asperger's work. Certainly, he was better known in English-speaking countries for years before the latter's papers were translated. The term 'Asperger syndrome' has since acquired a connotation that is far-removed from what Asperger himself described. It has become a sort of 'autism for the middle classes' and is about to be consigned to history. In the 5th revision of the American Psychiatric Association's Diagnostic and Statistical Manual, the eponym will be abolished. Lorna Wing, who was responsible for introducing Asperger's work to the English-speaking world (Wing, 1981), will not be sad to hear this. She has described its current usage as a 'political diagnosis' (personal communication).

The intense emotions that have been aroused over the years by arguments over how autism should be conceptualized, and its validity as a diagnosis in psychiatry, arose in particular from two contrasting theoretical positions. One had its origins in the writings of Bruno Bettelheim, whose 1967 book, The empty fortress: infantile autism and the birth of self, blamed parents for the development of their child's autism. Its psychoanalytical take on the origins of autism has, arguably, since been discredited insofar as no scientific evidence can be found to support the idea that parental behaviour causes autistic symptoms. Still, it is remarkable that in France, three quarters of psychiatrists still treat autism using a psychoanalytical approach, and much the same is true of Switzerland. There are places too in the UK, where NHS treatment for autism is delivered to (often non-verbal) autistic children within the same conceptual framework, inducing parental guilt as well as failing to deal satisfactorily with the key clinical and educational issues.

Another influential theory, espoused by Hans Asperger among others, was that autistic symptoms have strong continuity over time with the development of schizophrenia in early adulthood. The Journal of Autism and Childhood Schizophrenia,
published from 1971–79 under this hybrid title, until it became the Journal of Autism and Developmental Disorders. The apparent confusion was not helped by comments from Kanner, who wrote of his autistic subjects, ‘possibly all of them are schizophrenic’. The continuity of risk from early childhood autism to schizophrenia is not well understood. There are few longitudinal studies of the more able autistic children that have been increasingly diagnosed in recent years. Their social-cognitive difficulties resemble those of people with schizophrenia, in some respects. In contrast, most symptoms shown by moderate to severely mentally retarded people with autism, who represented the majority of recognized cases in the late 1970s, are not at all similar to those of schizophrenia. There is no doubt that idiopathic schizophrenia is not a common outcome of autism, although an incipient paranoia is not infrequently encountered in adolescents. The key point to bear in mind here is that the terms have really changed in their connotations over the past four decades. As the concept of schizophrenia has become more precisely delineated from other psychiatric phenomena (in a large part because of British research four decades ago) the definition of autism is also going through a major revision, towards a broader ‘spectrum disorder’ (DSM-5 Neurodevelopmental Disorders Working Group, 2011).

Our diagnostic practices are driven in part by cultural considerations; science is subject to cultural trends no less than other human pursuits. In the 1960s and 1970s an emphasis on the role of environmental influences on risk of mental illness was prominent. Even schizophrenia was thought to have its origins in family tensions, exemplified by the obscure, beautiful, but seriously misguided writings of Ronald David Laing (1960). Nowadays, impelled by technical advances in neuroscience and genetics, we are becoming ever more fascinated by the prospect of discovering the biological origins of mental disorders; and autism would seem to be an exemplar of that trend.

Exhibit number one, in the case of ‘genes versus environment’, as causes of autistic disorder, is the role played by genes as exemplified by the findings of twin studies (Ronald and Hoekstra, 2011). The early twin studies did indeed demonstrate that concordance for autism in monozygotic twins is far greater than the risk shared by fraternal twins. It is common to see a similar argument quoted by authors at the beginning of published papers on their genetic studies. They justify a search for phenotypic associations with single nucleotide polymorphisms on the basis that twin and family studies, undertaken over a period of several decades, have consistently shown that autism spectrum disorders have an overall heritability of about 90%. This statement seems to suggest that the potential role for environmental influences is very small (Skuse, 2007). The prevailing view is that autism risk is rooted in maldevelopment of the brain during infancy, and that this occurs because of faulty genetic programming. On the other hand, we also know that autistic-like symptom complexes can arise from early life circumstances if these fall well outside the boundaries of a reasonably normal environment. Circumstances of severe neglect, and paucity of interpersonal stimulation during infancy, especially individual attention from a caregiver, can be responsible. These situations are rare, but have been observed in impoverished orphanages (Rutter et al., 1999). An important conceptual issue arising from that observation is whether the apparently environmentally-induced cases of autism are sufficiently similar to ‘idiopathic’ cases to warrant the same diagnosis. This is not a trivial problem, in part because we do not know in detail just how much heterogeneity there is in the symptom profiles of the idiopathic syndrome. The reason for that lack of clarity is in part because of the remarkable constraints upon the research definition of autistic disorders. For a variety of reasons, such constraints have effectively precluded the scientific investigation of autism as comprising not one but potentially many related conditions.

Traditionally, autism has been regarded as having three main domains of impairment, although the term ‘triad’ can be misleading. Essentially, the diagnosis is made on the following basis: first, there are symptoms of impaired social reciprocity (the ability to engage in some ‘to and fro’ dialogue with another, the ability to see another person’s point of view). This impairment is critical and fundamental. Second, there is some abnormality in the use of language for social communication. This may amount, in its most extreme version, to mutism: affected individuals (with severe generalized learning difficulties in most cases) never speak, although their understanding of language is often rather better developed. Most people with an autistic disorder use language quite well in a formal sense, but it sounds odd and pedantic to others, and is sometimes hard to follow, lacking contextual cues and emotional inflexions. Finally, there is a set of symptoms that do not, on the face of it, have much in common at all, although they are grouped together. These include restricted interests (typically factual and memory-dependent), sensory sensitivities (especially to certain sounds, which can be distressing or distracting), motor stereotypes (for example, flapping hands when excited or anxious) and a pervasive resistance to change (inflexibility, possibly related to deficient executive functions). Diagnosis is by a combination of interview (with an informant) and observation of the individual’s behaviour. For research purposes, the diagnosis is made in the overwhelming majority of cases by a combination of copyrighted instruments [the Autism Diagnostic Interview (Lord et al., 1994), and the Autism Diagnostic Observation Schedule (Lord et al., 2000)], both of which were developed before the recent reappraisal of autism as a spectrum of disability.

If the risk of developing autism is so clearly related to a substantial genetic predisposition (which cannot have changed recently), then why do we see so many more cases nowadays in our clinics than we did a few decades ago? The first study of prevalence was done in the UK in 1966. For many years, a prevalence figure of four cases per 10 000 children was considered to be accurate. There were some important assumptions that went along with that figure. First, autistic symptoms were defined as being qualitatively distinct from typical behaviour. This strongly implied that such behaviours were not seen, or were very rare, in the general population of non-autistic children. Second, there was an assumption that autism was very closely linked to mental retardation, and that the same genes that predisposed to autism were risk factors for generalized learning difficulties. Third, there was a strong emphasis on language delay as the hallmark of early autism risk, and many thought that the condition was primarily a disorder of language development. Fourth, the concept of comorbidity received very little attention; if symptoms of another neurodevelopmental disorder (such as attention-deficit hyperactivity
disorder, Tourette syndrome or obsessive compulsive disorder) were observed, these were explicitly disallowed from being recorded separately but were assumed to be reflective of the same underlying pathology (Gargaro et al., 2011).

Within the past decade, we have been forced to reconsider our views of what autism is, and how it should be defined. This reconsideration is being driven, fundamentally, by the observation that autistic disorders are much more common than four per 10 000 children. Epidemiological studies consistently report figures of at least 25 per 10 000 for the most restrictively defined autistic disorders (often informally known as Kanner-type autism, because they fulfil Kanner’s essential diagnostic criteria). Beyond that core group, there is increasing evidence for a continuum of symptom severity in the general population (Skuse et al., 2009). The point at which we should create a cut-off between ‘clinically significant’ and ‘normal variation’ is unclear, but should be guided by evidence on functional impairment arising from symptom complexes, measured dimensionally. In the USA, over 1% of children (~2% of boys) have been given a diagnosis of some disorder on the autism spectrum (Kogan et al., 2007).

Virtually all the key assumptions that were held by the earliest generation of autism researchers, of whom many were interviewed for this book, are now being questioned. No wonder there is tension in the autism community. First, there is no qualitatively distinct symptom complex associated with autism that is not found in other children who lack any serious neurodevelopmental impairment (Happé et al., 2006). Second, there is no strong association with generalized learning difficulties; the proportion of identified cases with autism and low IQ is falling steadily (Baron-Cohen et al., 2009). Third, there is no inevitable delay in the onset of language, and the criterion will be dropped from the revised diagnostic framework currently being considered (DSM-5 Neurodevelopmental Disorders Working Party, 2011). Finally, comorbidity appears to be the rule rather than the exception, and it has become inevitable that separate diagnostic recording of other disorders such as attention-deficit hyperactivity disorder is introduced, because it makes clinical sense.

In his History of autism, Adam Feinstein has documented in an entertaining and yet exacting way, the evolving history of how our present-day ideas about autism reflect the personalities and interests of key individuals, who played critical roles in our understanding of this perplexing symptom complex. He has been to interview them, all over the world, and draws some fascinating responses to his questions. It is particularly pleasing that he managed to gather the material for his review before the most elderly among the pioneers passed away, as several have done within the past 2 or 3 years. Adam Feinstein has not set out to write a revolutionary account of the subject. Somehow, given the intensity of the debate engendered by the personalities involved, he has managed to side-step many of the most bitter controversies. Still, he does reflect the current fractured state of affairs. He gathered his history in cities from London to Melbourne, and makes a fine job of summarizing his findings, although he wisely refrains from drawing conclusions.

In her interview with Adam Feinstein, Lorna Wing (to my mind, one of the most provocative and insightful of his interviewees) told him Leo Kanner was convinced that autistic children had a potential for normal or high intelligence. The idea that autistic people have hidden talents has of course been reinforced by the popular depiction of characters such as Rain Man and the idea has gained currency that several brilliant artists and scientists have had Asperger syndrome too. This notion, of a savant syndrome attracts tremendous attention, especially when some researchers claim that the proportion of people with autism who have savant skills could be as high as 30% (Howlin et al., 2009). Perhaps the leading expert on this subject nowadays is Dr Darold Treffert, of Fond du Lac, Wisconsin. He has recently authored a biographical review of savant skills in autism and other conditions (Treffert, 2010). The film Rain Man was released in 1988, starring Dustin Hoffman, who had made a close study of individuals with high functioning autism. Darold Treffert was one of the key advisors on the film, and he was happy that it provided a positive depiction of autism that would be, on the whole, beneficial. The screenwriter was inspired by the true story of Kim Peek, who had a phenomenal memory and could recall the exact contents of over 12 000 books. Most readers who know anything about a savant syndrome in autism will have learned about it from Hoffman’s Oscar-winning performance.

When we consider the Islands of genius, described in the book of that name, these islands seem not only to be observed among those individuals we regard as socially disabled with autistic disorders. Rarely, they may be acquired, perhaps after a head injury. Quite how that happens is still unknown, but it makes for a fascinating narrative. True savant skills are so far removed from the normal range of abilities that they seem almost miraculous. I once had the opportunity of observing a remarkable savant, who never became as celebrated as the characters in this biography for the simple reason that his parents wished to protect him. At the age of a little over 4 years, with no spoken language, he was able to derive cube roots from 7-digit numbers using mental arithmetic and writing the resulting number from right to left (that is, with the last decimal place first). He could also type, with two fingers, in a way that allowed us to estimate his superior verbal IQ. He could write moving passages of self-description, with apparent insight into his condition, in this way. There was no ‘facilitated communication’ involved. Unsurprisingly, his nursery school thought he was mentally retarded and probably ineducable. To all intents and purposes, he appeared utterly autistic.

Darold Treffert divides savant skills into three main levels of ability. First, there are splinter skills, which are typified by an obsession with trivia, found in many people with autism spectrum disorders. The key to this skill seems to be the combination of a good rote memory and over-practice. We once saw a young man, about 14 years old, who had rather low general abilities but had a great interest in bus routes. He was so infallibly talented in his recall that the local bus garage used him to train new drivers. He was amply rewarded with CD players, bicycles and other adolescent desiderata. The second set of skills in this classification provide for limited talents, such as musical or other artistic abilities, often in a single area. Some children with autism can develop these skills, with support, to a high degree. Interestingly, the harnessing of their talent may lead to a growth in self-esteem and...
better social adjustment. Thirdly, there are a few people with what Treffert describes as ‘prodigious abilities’. These are abilities that would stand out in anyone, because they are remarkably well developed against a background of generalized limitations in other areas. An example might be the acclaimed autistic artist, Stephen Wiltshire, who took a 15-min helicopter ride over London and then spent 5 days drawing an amazingly detailed view of 7 square miles of the city.

No single theory can explain all savant skills, but there are common themes. First, most savants have a remarkable memory, which can be in the domains of the visual arts, music (e.g. 7000 compositions played by ear), calculations, calendars or spatial ability. Occasionally, it is a rather banal ability to remember in detail events one has experienced, even the entire history of one’s own life. Usually, the savant skills tend to be isolated, so there is rarely a remarkable talent in more than one area. The investigation of such skills has resisted attempts at scientific explanation. Some have tried to discover how a prodigious but eccentric ability develops, such as calendar calculating skill, by means of functional neuroimaging. The phenomenon resists any simple explanation, although it seems intensive practice is almost certainly a key feature.

Perhaps the most controversial and puzzling aspect of savant skills is the evidence that rare individuals did not have any outstanding ability prior to an event that caused brain trauma, but that their talent developed after the trauma, or after the onset of some degenerative process such as dementia. Subsequently, they acquired a remarkable facility, often associated with a prodigious memory. Many parents of severely autistic children believe that their child has dormant skills. If only they could be released, hitherto concealed talents and communication abilities would be revealed. The evidence that there is an ‘inner child’ in cases of autism is slim. On the other hand, Treffert has identified a handful of otherwise typical children and adults where history suggests that certain remarkable abilities (both creative and computational) only became apparent after an event such as a blow to the head, meningitis or progressive brain disease. He takes the view that we all potentially possess an ‘inner savant’ inhabiting our right cerebrum. In his opinion, our non-dominant cerebrum is a key feature.

References


