Autism and specific language impairment: categorical distinction or continuum?

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Abstract. Traditionally, autism and specific language impairment (SLI) are regarded as distinct disorders, with differential diagnosis hinging on two features. First, in SLI one sees isolated language impairments in the context of otherwise normal development, whereas in autism a triad of impairments is seen, affecting communication, social interaction and behavioural repertoire. Second, there are different communication problems in these two conditions. Children with SLI have particular difficulty with structural aspects of language (phonology and syntax). In contrast, abnormal use of language (pragmatics) is the most striking feature of autism. However, recently, this conventional view has been challenged on three counts. First, children with autism have structural language impairments similar to those in SLI. Second, some children have symptoms intermediate between autism and SLI. Third, there is a high rate of language impairments in relatives of people with autism, suggesting aetiological continuities between SLI and autism. One interpretation of these findings is to regard autism as ‘SLI plus’, i.e. to assume that the only factor differentiating the disorders is the presence of additional impairments in autism. It is suggested that a more plausible interpretation is to regard structural and pragmatic language impairments as correlated but separable consequences of common underlying risk factors.

Language impairment is a central feature of autistic disorder. The 1970s saw researchers focusing on whether language was the central feature of autism, i.e. asking whether the other symptoms of this disorder were secondary consequences of limited language skills. To this end, comparisons were made between children with autism and those with specific language impairment (SLI). The answer seemed clear-cut: the syndrome of autism could not be attributed to language difficulties: symptoms were more severe, more extensive, and different in kind from those seen in SLI. Consequently, contemporary diagnostic frameworks draw a sharp dividing line between autism and SLI and emphasize the differential
diagnosis of these conditions. Nevertheless, in recent years, this neat division has been questioned. Cases have been described who show an intermediate clinical picture. Furthermore, family studies have suggested possible aetiological overlap between SLI and autism. In this chapter, I review this recent evidence, and consider the implications for studies of the aetiology of developmental disorders.

Language and communication in SLI and autism: the conventional view

SLI is defined when a child fails to acquire language at the normal rate for no apparent reason. Non-verbal ability is within normal limits, and there is no indication of physical or sensory handicaps that could account for the language difficulties. Although it is widely accepted that SLI is heterogeneous (Bishop 1997), for most children the principal difficulties are with structural aspects of language, i.e. mastery of phonology (speech sounds) and syntax. It is usually assumed that children with SLI have normal non-verbal communication and social use of language—or if there are problems, these are simply secondary consequences of the structural language difficulties.

The language abilities of children with autism vary tremendously. Around 50% do not learn to talk and have severe comprehension problems. Others acquire language late and do not progress beyond simplified speech. Distinctive features of autistic language are most readily observed in children of normal nonverbal ability—cases of so-called high-functioning autism (HFA). Many of these do acquire speech and may talk in long and complex sentences. However, their use of language is abnormal (see Table 1). Lord & Paul (1997) noted that whereas in SLI, children who talk most tend to be the most competent communicators, in autism, it is often the most talkative children in whom communicative abnormalities are especially apparent. Problems in the appropriate use of language in context come under the domain of pragmatics, and are the most striking feature in autism. On the basis of these contrasting phenotypes, SLI and autism are usually thought of as distinct disorders with different aetiologies, as illustrated in model A (see Fig. 1).

Comparisons of communication in SLI and HFA

Although the textbook accounts of these disorders suggest a clear divide, some studies suggest that the boundaries between these disorders are not so sharp. On the one hand, on standardized tests of structural language skills, children with HFA often have deficits similar to those seen in SLI. On the other, there is evidence of pragmatic difficulties in some non-autistic children with language impairments.
**TABLE 1** Typical characteristics of language and communication in verbal children with autism (based on Lord & Paul 1997)

<table>
<thead>
<tr>
<th>Characteristic</th>
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<tbody>
<tr>
<td>First words acquired late</td>
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<tr>
<td>Marked impairment in language comprehension</td>
</tr>
<tr>
<td>Articulation normal or even precocious</td>
</tr>
<tr>
<td>Abnormal use of words and phrases with idiosyncratic meanings</td>
</tr>
<tr>
<td>Use of made-up words (neologisms)</td>
</tr>
<tr>
<td>Pedantic and over-precise speech</td>
</tr>
<tr>
<td>Dissociation between mastery of grammar and functional use of language</td>
</tr>
<tr>
<td>Echolalia</td>
</tr>
<tr>
<td>Confusion and interchanging of personal pronouns, such as I/you</td>
</tr>
<tr>
<td>Abnormal vocal quality</td>
</tr>
<tr>
<td>Abnormal intonation and stress</td>
</tr>
<tr>
<td>Failure to use contextual information in comprehension</td>
</tr>
<tr>
<td>Over-literal interpretation without appreciation of speaker’s intention</td>
</tr>
<tr>
<td>Low rate of spontaneous initiation of communication</td>
</tr>
<tr>
<td>Little reference to mental states</td>
</tr>
<tr>
<td>Persistent questioning</td>
</tr>
<tr>
<td>Poor at judging what a listener needs to be told</td>
</tr>
<tr>
<td>Difficulty in making causal statements</td>
</tr>
<tr>
<td>Lack of cohesion</td>
</tr>
<tr>
<td>One-sided talk rather than to-and-fro conversation</td>
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**MODEL A**

FIG. 1. Model A: distinct causal routes for SLI and autism. Both disorders are highly heritable, but it is assumed that different sets of genetic risk factors are implicated in the two disorders.
Sigman & Capps (1997) summarized the communicative features of autism by concluding that phonological, semantic, and grammatical development tend to follow a normal course (albeit at a slow rate in children with low IQ), but language use does not, and is aberrant. Nevertheless, in most children with autism, structural language skills are at least as poor, if not worse, than those of children with SLI. Figure 2 shows illustrative test data from Lincoln et al (1993). In a much larger study, Fein et al (1996) compared language scores of preschool children of normal non-verbal ability who had autism or SLI. Although profiles of language scores were more uneven for the children with autism, on no test did they significantly outperform children with SLI. Kjelgaard & Tager-Flusberg (2001) did not directly compare autism and SLI, but they used a broad range of language measures with a large group of children with autism. They found that in general these children had impaired expressive and receptive language, and there was a clear relationship between IQ level and language skills. Articulation skills were almost always unimpaired, but on a test of non-word repetition, in which the child repeats back meaningless strings of sounds such as ‘blonterstaping’, many children with autism did very poorly. They noted that poor non-word repetition in children is frequently seen in SLI (e.g. Bishop et al 1996). Overall, then, studies using standardized language measures suggest children with autism have many of the same impairments as are seen in SLI: these, however, tend to be overlooked because the pragmatic difficulties are more severe and unusual.

Evidence for cases intermediate between SLI and autism

A landmark study in this field was initiated by Bartak et al (1975). They recruited boys aged 4.5–9 years who had broadly normal non-verbal IQ but severe comprehension problems, and found that most of them could be categorized as cases of autism or receptive SLI (‘developmental dysphasia’). A detailed psychometric assessment was carried out, together with a parental interview. The main conclusion was that children with autism have distinctive pragmatic difficulties not seen in SLI (though, as in the studies reviewed above, structural language impairments similar to those in SLI were also present). Nevertheless, five of the 47 children recruited to the study could not be unambiguously classified in either category: their symptoms were intermediate and tended to change with age. Furthermore, when the sample was followed up in middle childhood (Cantwell et al 1989) and adulthood (Howlin et al 2000), the distinction between groups became blurred. Many cases from the language-impaired group developed autistic-like symptoms in non-language domains. This study suggested that the boundaries between autism and SLI might be less clear-cut than originally thought.
The same conclusion was suggested by other research on subtypes of language impairment. Rapin & Allen (1983) coined the term ‘semantic pragmatic deficit syndrome’ to refer to children who used fluent and complex language, but had abnormalities of language use, producing tangential or irrelevant utterances. Bishop (2000), who described similar cases, suggested the term ‘pragmatic language impairment’ (PLI) is preferable. The diagnostic status of these children has been the matter of some debate, because their language difficulties are reminiscent of those in HFA, yet, according to both Rapin & Allen (1983) and Bishop (1998) this language profile can be seen in children who are sociable and do not show major autistic symptomatology. Bishop & Norbury (2002) used standardized autism diagnostic instruments that assessed both current status and past history with a group of children recruited from special schools for those with communication impairments. None had a definite diagnosis of autism, though some had been described as ‘on the autistic spectrum’. Twenty-eight children had evidence of pragmatic difficulties on the Children’s Communication Checklist
(Bishop 1998), and were designated as the PLI group. The remaining 17 children did not have evidence of pragmatic difficulties and formed the typical SLI group. Table 2 shows how these children scored in relation to cut-offs for autism and its milder variant, Pervasive Developmental Disorder Not Otherwise Specified (PDDNOS) on both a parent report measure and direct observation. Although five of the children with PLI met diagnostic criteria for autism on both measures, the majority had autistic features that were not pervasive or severe enough to merit a diagnosis of autism. Six cases had no significant autistic symptomatology on either parental report or direct observation. There was also evidence that children’s symptoms changed with age, with autistic symptoms declining as they matured. An unexpected finding from this study was the relatively high rate of autistic symptomatology seen in the SLI-T group on the parental report measures.

Family studies and the ‘broader phenotype’

Superficial similarities between autism and SLI do not on their own provide particularly convincing evidence for common origins. After all, the same pattern of behaviour can arise for different reasons. However, evidence that the symptom overlap may reflect deeper commonalities comes from studies of relatives of people with autism. Although relatives meeting diagnostic criteria for autism are rare,

**TABLE 2** Numbers of children with typical SLI and pragmatic language impairment (PLI) categorized according to parental report and direct observation\(^{a,b}\)

<table>
<thead>
<tr>
<th>Diagnosis from parental report</th>
<th>Unaffected</th>
<th>PDDNOS</th>
<th>Autism</th>
</tr>
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<tbody>
<tr>
<td>Unaffected</td>
<td>7 SLI</td>
<td>5 SLI</td>
<td>2 SLI</td>
</tr>
<tr>
<td></td>
<td>6 PLI</td>
<td>4 PLI</td>
<td>4 PLI</td>
</tr>
<tr>
<td>Diagnosis from direct observation</td>
<td>PDDNOS</td>
<td>SLI</td>
<td>1 SLI</td>
</tr>
<tr>
<td></td>
<td>3 PLI</td>
<td></td>
<td>2 PLI</td>
</tr>
<tr>
<td>Autism</td>
<td>1 SLI</td>
<td>2 PLI</td>
<td>5 PLI</td>
</tr>
<tr>
<td></td>
<td>2 PLI</td>
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\(^a\)The Autism Diagnostic Interview — Revised (Lord et al 1994) or the Social Communication Questionnaire (Berument et al 1999) were used to obtain parental report of autistic symptoms, focusing largely on the period when the child was aged 4–5 years. These two measures gave good diagnostic agreement. The Autism Diagnostic Observation Schedule—Generic (Lord et al 2000) was used for diagnosis based on observation of current behaviour.

\(^b\)From Bishop & Norbury (2002) studies 1 and 2 combined.
cases of subthreshold symptomatology are common, including people who have linguistic and communicative difficulties resembling SLI and/or PLI (Bolton et al 1994). In the only large-scale study that directly compared family histories of children with autism and those with SLI, Rapin (1996) reported that rates of siblings affected with SLI were as high for children with autism as they were for those with SLI (see Table 3).

**Autism as ‘SLI plus’**

The overlap between autistic and SLI symptomatology, both within individuals and within families, raises questions about model A as an accurate depiction of the relationship between these disorders. An alternative would be to treat these disorders as points on a continuum of severity: mildly impaired cases have only structural language problems, and more severely impaired people have structural and pragmatic impairments, often accompanied by non-linguistic symptoms of autism. This simple view can readily be rejected, because it predicts that the most severe structural language problems should be seen in those with pragmatic

<table>
<thead>
<tr>
<th>TABLE 3 Rates of SLI and autism in first degree relatives of children with SLI or autism</th>
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<tbody>
<tr>
<td>Child diagnosis³</td>
</tr>
<tr>
<td>SLI ( (n = 192) )</td>
</tr>
<tr>
<td>% with an affected immediate family member</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>% with affected parent</td>
</tr>
<tr>
<td></td>
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<tr>
<td>% with affected siblingᵇ</td>
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-Based on Rapin (1996).
³Rapin (1996) used conventional diagnostic criteria for SLI, but preferred the term ‘developmental language disorder’ to refer to this group.
ᵇExcludes 10% of SLI, 39% of high-functioning autistic, 27% of low-functioning autistic and 26% of non-autistic control group who had no siblings.
difficulties, and this is not the case, either for those with autism or with PLI (Bishop 2000). Indeed, some children with HFA score within normal limits on structural language tests (Kjelgaard & Tager-Flusberg 2000). Thus though structural and pragmatic language difficulties tend to co-occur, there can be double dissociation—some have pragmatics unaffected and poor structure, and others have structure unaffected and poor pragmatics. This indicates that one deficit is not logically dependent upon the other, and implies distinct neurological bases for these aspects of communication. Model B (Fig. 3) depicts one way of accounting for this pattern, in which autistic disorder involves multiple underlying impairments, each with its own cause. According to this model, there might be a range of genetic risk factors, each of which affects a different brain system and leads to a specific set of symptoms. Autism would result when a child
was unfortunate enough to inherit a particularly disadvantageous constellation of alleles (W, X, Y and Z), leading to the full clinical picture. Model B is compatible with the heterogeneous symptomatology seen in developmental disorders affecting communication: for instance, it is possible to have cases of Asperger syndrome with good structural language skills, in whom only X, Y and Z are implicated, or relatively pure cases of PLI, where only risk allele X is present. On this model, the familial association between SLI and autism is explained by assuming that the pathway from genotype W to structural language impairment is usually implicated in autism.

Reconceptualizing the relationship between autism and SLI in this way would have major consequences for how we conduct genetic studies. Rather than working with a phenotype defined in categorical, clinical terms, we would move to a more dimensional approach, in which we look for quantitative trait loci linked to different aspects of autism, including language structure and use. An advantage of this approach is that we could assess these traits in parents and other relatives, rather than having to rely on multiplex families with more than one child who meets full diagnostic criteria for autism. Furthermore, model B implies that, in studies of autism, rather than relying on conventional standardized language tests, we should use language measures that have been shown to be sensitive phenotypic markers of heritable SLI, such as non-word repetition and tests of tense marking (Rice 2000).

Model B is attractive, in that it implies we might see clearer genotype-phenotype relationships if we move to a quantitative, dimensional view of autism. However, before we become too enthused with such a model, it is important to note several arguments against it. First, there are phenotypic differences between SLI and autism that raise difficulties for this account. As noted above, children with autism usually do well on articulation tests, whereas those with SLI often make errors in producing speech sounds, despite normal motor control of the articulators—i.e. they have a phonological impairment. This, then, is one aspect of communication where one cannot make the generalization that children with autism have the same deficits as SLI.

Second, the model entails that the different domains of impairment in autism have independent origins, and their co-occurrence in autism is a matter of chance. For this to be the case, the risk alleles implicated in genotypes W, X, Y and Z would need to be very frequent in the population, and cases of isolated symptoms should be extremely common. We do not have epidemiological figures on rates of autistic symptomatology in the general population, or on co-occurrence of symptoms, but it seems unlikely that symptoms are independent of one another.

Third, different symptoms tend to co-occur within families. If the combination of risk alleles determined the pattern of symptoms, then monozygotic (MZ) twins
should be phenotypically identical for autistic symptoms. LeCouteur et al (1996) found this was not so: variation between two members of a MZ twin pair was as great as that seen between twin pairs. Clearly some factor other than genetic makeup influences the symptom profile. Furthermore, phonological problems, which are not usually part of the autism phenotype, are seen in relatives of people with autism.

The final argument is based solely on precedent—in general, single gene disorders that affect brain development do not influence a discrete brain system and cause a distinctive modular impairment. Although it is often possible to identify a prototypical behavioural phenotype associated with a genotype, there is often substantial individual variation within a given genetic syndrome. Neurofibromatosis type I is an instructive example (Reiss & Denckla 1996): the same genetic mutation can affect different brain regions, and lead to very different clinical pictures, e.g. a parent with mild symptoms may have a severely affected child. To complicate matters even further, there are also ample instances of different genes leading to the same phenotypic outcome, e.g. phenotypically identical manifestations of tuberous sclerosis are caused by genetic mutations on chromosomes 9 and 16 (Udwin & Dennis 1995).

Language impairment and autistic disorder associated through pleiotropy

Such considerations suggest we need to consider an alternative account, model C (Fig. 4), in which autism and SLI have at least partially distinct neurological bases, but common aetiological factors affect both of them. For instance, suppose that there are genes that disrupt processes of neuronal migration, leading to abnormal brain structure. The precise outcome of such a process will depend on which brain systems are implicated, and this might be affected by the genetic background (i.e. other genes interacting with the risk genes), systematic environmental influences, or chance events.

This model differs from model B in that it predicts that some cases of SLI will have symptoms that go beyond what is seen in autism. It can accommodate the apparently paradoxical finding that symptoms that frequently go together need not necessarily do so, by assuming that different symptoms have different brain bases, but common aetiological factors can disrupt their neurodevelopment. However, although this model may be plausible, it suggests that the enterprise of discovering the genetic basis of autism is going to be considerably more difficult than we might have imagined. It also raises questions about where we draw the boundaries of the phenotype. On the basis of both family and behavioural data, I have argued for continuities between language impairment and autism, but one could ask why stop there? There is considerable comorbidity between SLI and attention deficit disorder (Beitchman et al 1996), literacy impairments (McArthur
FIG. 4. Model C: a range of genetic risk factors (X, Y, Z, etc) is implicated in the aetiology of developmental disorders, each of which can affect separate brain systems: the dotted path from genotype to brain systems indicates a probabilistic influence. The phenotype will depend on which systems are affected, and this could be a function of genetic background, systematic environmental influences, or random factors. Correlations between symptoms may reflect involvement of adjacent brain systems.

et al 2000) and motor immaturity (Bishop 2002). Are these also influenced by the same genes that pose a risk for autism?

Model C implies that two kinds of study need to be at the top of our research agenda. First, we need behaviour genetic studies that can clarify which developmental disorders are heritable, and how far comorbid traits are influenced by the same genes (Rutter 2000). Such studies depend on the availability of reliable, quantitative methods of assessment that can act as sensitive markers of underlying genotype. Second, studies of MZ twins teach us that, even though autism is a
strongly genetic disorder, non-genetic factors are important in determining the phenotype. Consider the MZ twin pair described by Rapin (1996), one of whom had classic autism and the other SLI. The difference may be due to chance influences early in development that determine which brain regions are subject to genetic effects, but before accepting that, we need to look for more systematic environmental influences that may play a part. Within-family comparisons of affected relatives, and contrasts between children who follow different developmental courses have the potential to throw light on environmental factors that may influence severity and pattern of autistic symptomatology in genetically at risk individuals.

Conclusions

Autistic disorder and SLI have traditionally been regarded as distinct disorders, but recent work suggests some overlap both at the phenotypic and the aetiological level. One way forward would be to view autism as a form of SLI in which a broader range of impairments is present, and to look for genetic correlates of specific components of the autistic triad. However, we need to be cautious about assuming a simple one-to-one relation between genotype and phenotype: it is more likely that there are genetic risk factors that have the potential to compromise brain development, but their precise impact depends on the genetic background, environmental influences and chance factors, sometimes leading to SLI, sometimes to autism, and sometimes to an intermediate clinical picture. The answer to the question, are autism and SLI on a continuum, depends on the level of description. Phenotypically the pragmatic communication deficits seen in autism are not continuous with structural language impairment: they can show a pattern of double dissociation, indicating they are logically separable. However, aetiologically, they appear to share common risk factors.

Acknowledgements

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References


DISCUSSION

Fombonne: I was interested in your comments that perhaps you are seeing fewer diagnoses of SLI. Do you have evidence for this?

Bishop: We need to do a study on this. I don't think this is just a case of diagnostic labels: it is more my impression of the children I find when I go into schools. When I did my PhD I saw 80 children, and about 10 of these looked as if they had obvious pragmatic problems. Now when I go to schools it is very hard to find children with classic SLI who don't have some sort of syndrome. It could just be a case of who is getting into which schools. If it is the case that rates of autistic spectrum disorder are increasing, it could be that those children are pushing out the classic SLI cases, who therefore end up in mainstream schools instead. However, I'm intrigued by another fascinating possibility, namely that autism and SLI may involve the same core disorder and it is just the way it is manifesting that has changed.

Fombonne: In your ADI scores, in the autistic upper right corner, with the children with SLI diagnosed with autism by the ADI, were the scores very high like in autism samples, or were they just meeting criteria?

Bishop: In general most of these children were just meeting criteria. We are talking about children who are coping in a special school which didn't have particular facilities for dealing with massive behavioural problems. What was low in this sample was repetitive behaviour of any severity.

Fombonne: If you start with SLI problems or pragmatic syndromes, have you looked at the family data in terms of rates of autism?

Bishop: We do have some of these data but I haven't analysed them yet. There are a couple of children in this sample with pragmatic problems who have older siblings with classic autism.

U. Frith: To my mind there is a pressing need to study variable or discordant cases, either in twins or in multiplex families. It is extremely important to verify
how different they really are. One possible outcome is that these cases look very different on the surface, but are not essentially different in terms of neurocognitive deficits. The alternative outcome, namely that there are differences in the basic neurocognitive deficits even in genetically related cases, is extremely interesting. Susan Folstein gave a hint of this possibility. She analysed different items on the ADI and showed cross correlations on different items between two affected siblings. Mostly these were high. However, there was one dimension where there was no cross-correlation on ADI measures between affected siblings. Strangely enough, this was in the social communication scale. This needs to be followed up, because after all social communication impairment is core to autism. Could it be that as currently assessed on the ADI it is less subject to genetic influence than other signs of autism? If we measured social impairment in different ways (for example by laboratory tests) would the same lack of cross-correlation be obtained?

Bishop: I take your point in general. Moving away from whether or not subjects meet criteria, to exploring to what extent these symptoms are similar or different within children, is absolutely key. We have only just begun to do this. The other issue that concerns me is whether it is appropriate to be looking at the symptom level, and whether we shouldn't perhaps be using some of these measures of underlying processes such as Francesca Happé was talking about. My worry is that I am not convinced that we have good measures with adequate psychometric properties.

Folstein: I also mentioned that both Peter Szatmari and Jeremy Silverman, using two separate autism data sets found the same thing: most of the ADI components do have sib–sib correlations, but not social intent.

Rutter: What do you conclude from that?

Folstein: First I thought it was related to the birth order severity effect. There are, however, differences in the age at which the ADI was done. Older children were given more severe ratings by their parents. I don't know whether that has something to do with optimism or different interventions in different cohorts. Another thing that I thought is that social interaction is the sum of a lot of different parts. It is not a separate entity. I also feel that when I see the parents, sometimes. They are unsociable for several different reasons, or unsuccessful in their sociability, rather than intending to be unsociable.

Fombonne: There might be some contrast effects, as well. If the same informants report on two different children of their own, they might artificially increase the contrast between the two, as has been shown in twin studies.

U. Frith: In this case you should also find low correlations in the other dimensions.

Lord: Not necessarily. In the ADI, the codes for the social questions are more clearly pegged to the questions. Frequently, in the last sections of the ADI, the
examiner asks the parent whether the child ever behaved in a certain way, and the parent responds by describing a behaviour that is coded under other headings. If you ask, 'Does your child have any usual interests?' The parents might say, 'Yes, he spins everything that he sees'. I wonder if there you might get more similarity, not in terms of what the parents say but in terms of the examiner who codes it, than in social where the code is more clearly prescribed.

Bishop: I was very surprised that people were not reporting or making use of the data from the ADI on current functioning as opposed to earlier functioning. We re-scored our ADI data in terms of what subjects are like now rather than what they were like in the preschool period. It was clear that a lot of the children improved quite markedly over time. This might be a useful thing to do more generally. Because autism is regarded as a lifelong disorder, once it is diagnosed people seem to lose interest in how symptoms may change. But certainly for these marginal children they can change a lot.

Folstein: When we just put in the 'ever' codes there was not enough variation in the coding to get any sensible factors. Most autistic subjects have most of the symptoms at one time or another, which would be coded as 'ever'. We didn’t want just to put the current ones in, because somehow this didn’t seem to give a good view, so we put both in.

Bishop: I’m thinking merely in terms of documenting the natural history of this disorder. I would have liked to know how our children compared with others in terms of changes over time. I could not find anything in the literature on this.

Sigman: We have looked at change. We see stability in the low-functioning children, and parents report decreased severity of symptoms in the high-functioning children.

Buitelaar: Might Susan Folstein’s finding of an absent sibling correlation for social deficit scores be an artefact due to the fact that both siblings have high deficit scores in the social domain, and that there is reduced variability?

Folstein: There is a broad distribution of the scores on the social factor.

Rutter: You would have to postulate that you don’t have that methodological problem with the other symptoms. I would be surprised if that was the case.

Bailey: It is worth adding that in the IMG SAC sample we have exactly the same finding: there is no familial clustering in the ADI social domain, but there is clustering in the non-verbal communication and repetitive domains. This is correcting for IQ and age when the ADI was carried out.

Dawson: It is a question of variability. We would anticipate more variability in general in language and repetitive behaviours.

Bailey: No. The possible ADI score is much higher in the social domain.
Folstein: There are more items there, but what I did was to make them all come out to a maximum of 1, to account for the fact that different numbers of items were loaded on different factors. This wasn't an issue in our analysis.

Rutter: Chris Hollis, could you say something about Judy Clegg's further follow-up under your supervision?

Hollis: This adds to Francesca Happé's discussion about the similarities and differences between cognitive processes involved in autism and SLI. We continued Mike Rutter and Lyn Mawhood's follow-up of the receptive SLI group into their mid-30s. Rutter and Mawhood had previously found that the SLI group had quite marked and unexpected social impairments in their early 20s. We had two contrasting groups: an IQ matched control group and siblings without a history of language disorder. We assessed them in terms of their social function, language and literacy, phonological processing ability (non-word repetition) and three different measures of theory of mind. The SLI group had significant impairments on both phonological processing and theory of mind measures—but these two domains of impairment were not correlated. This finding suggests some independence between on the one hand, phonological processing, language and literacy and on the other, theory of mind, social cognition and social functioning.

Bishop: I'd be happy with that on the basis of my viewpoint.

Hollis: For various reasons we didn't re-assess the autism group. So, I am interested in whether cognitive measures of language processing, such as non-word repetition are abnormal in autism.

Bishop: Kjelgaard & Tager-Flusberg (2001) did that and found that many of the children with autism were very poor at non-word repetition. If you looked at the group as a whole on average they performed very poorly, but there were still some with autism who were doing fine. Nicola Botting and Gina Conti-Ramsden have found the same with pragmatically impaired children (Botting & Conti-Ramsden 2002): some are severely impaired and some score within normal limits on non-word repetition. It is a messy picture. There are variable symptoms that occur probabilistically without seeming to have very strong causal links to one another.

Rutter: You have a fascinating set of data and a persuasive model. What I find really puzzling, though, is why this language group was so relatively normal in their social behaviour early on, and yet the individuals developed quite marked problems later. Whether measured psychometrically, socially or behaviourally, this was the case. Why so late?

Bishop: One answer could be that this is not like autism at all and that this is the consequence of being stuck in the big wide world not understanding much of what is going on around you, in a rather unsympathetic environment. It may be that this is a symptom whose manifestation depends on the intervention that the children receive. The reason I have tended not to favour this explanation is that children
with profound deafness don't seem to end up looking like children with autism, despite their poor understanding of oral language. Of course, many deaf children have exposure to a rich language community through sign language, but that is not always true in the UK. Some deaf children are stranded in mainstream schools without other deaf people around them, yet they do not become autistic. We tend to regard social impairment as a consequence of poor understanding, but if you interact with deaf children you find that they are socially so normal.

_Hollis:_ I don't think the social difficulties seen in some children with SLI can just be explained as a consequence of their language problems. The finding of increasing social difficulties while language function improves over time argues against this. We matched the SLI group with controls with equivalent IQ and found the controls were functioning socially far better. We then compared the SLI group with a performance-IQ matched sample from the National Child Development Cohort (NCDS). Again, the SLI group were functioning much worse than IQ matched controls. While at one level this may look like evidence for a possible causal link between language and social impairments, the underlying cognitive mechanisms involving phonological processing and social cognition appear to be independent. This suggests that both types of cognitive deficit may be required to produce social impairment and SLI, whereas a specific phonological processing deficit may result in SLI or dyslexia without significant social impairment.

_Bishop:_ You said you matched on IQ. Is that the same as language functioning? Was this verbal IQ?

_Hollis:_ They were matched on performance IQ.

_Bishop:_ When you were conversing with them, were they able to understand at speed and formulate language at speed in a social interaction as fluently as other people? I do think sometimes that children can look all right in the test situation but still not be able to perform so well in everyday life when they are under time pressure.

_Rutter:_ Very few of these adults with a developmental language disorder would be regarded as showing autism. Their social behaviour was closer to autism than was the case when they were young but the groups with autism and specific language impairment continued to be different in important ways (Howlin et al 2000).

_Charman:_ What are the most important differences between an adult high-functioning autistic sample and SLI?

_Rutter:_ There is less in the way of repetitive and stereotyped behaviour with SLI. In terms of language, as in the earlier follow-up, there was less language abnormality as distinct from poor communication.

_Bishop:_ What is their non-verbal communication like?
Hollis: It is difficult. They weren’t initially selected as being a pragmatic-impairment group, so probably not all of them would also fit your pragmatic-impaired group.

Bishop: Certainly, within our pragmatically impaired group we see some who have dreadful non-verbal communication as well. We have some who have good eye contact and ability to use facial expression: they look very normal non-verbally, but they come out with odd things and use rather stereotyped and odd intonation that are classic for high-functioning autism.

Monaco: With regard to the late-onset of the systems in the pragmatically impaired group, could you not look at the families and use variance component modelling to get at whether this is ‘environmentally’ induced or is primarily genetic? If you are going to break the measures into some kind of distribution, you can then compare this with the variance of the siblings and attribute this to environmental or genetic causes.

Bishop: The trouble is, it could be genetic and late-onset.

Monaco: But if they are purely environmental it will come out.

Bishop: I don’t see how this relates to early or late onset. It would just tell us whether things are heritable.

Folstein: Helen Tager-Flusberg’s sample has been alluded to. I have been involved in this study, and one of my roles was to do a psychiatric interview with the parents. I was struck by how often the children with SLI had social phobias that were particularly related to speaking in public, such as talking on the telephone or asking a stranger for directions, even after they had acquired quite adequate language capabilities. Their mothers would say that for so long they were unable to speak well that they began to avoid it and became fearful of it. On the other hand, if they had to be in a play they weren’t as bad because then they memorised and practised their lines. Their problem is with spontaneous speech. Helen and I are now doing a study comparing the language phenotypes — including pragmatic language — of the parents and one randomly chosen sibling in autism and SLI families. The probands are matched on verbal IQ. We still have children in the old ‘mixed’ language/autism group.

Rutter: While we are discussing age manifestation differences, can anyone help explain the findings in our study of Romanian adoptees. In the published paper (Rutter et al 1999) we compared them with one of Cathy Lord’s longitudinal studies. The two groups were indistinguishable on the ADI at age 4 but they were already different at 6 years. We are now about three-quarters of the way through a further follow-up at age 11. The findings so far suggest appreciable further change. The circumscribed interests that were so striking early on have gone completely in some cases, and have faded in the majority. The language abnormalities are also much less evident. With some important exceptions, their social behaviour would no longer be regarded as autistic-like. The children have
plenty of social problems, but they seem to have more in common with disinhibited attachment. I am as puzzled over why the autistic-like behaviour in this group diminished with age, as I am with the increase with age in such behaviour in the SLI group.

Skuse: We have to come back to the notion of a sensitive period in early postnatal life, during which an experience that would normally have occurred didn’t occur. There is no reason to think that this particular group was genetically predisposed in any way to be autistic, and they weren’t abandoned there because they were autistic-like.

Rutter: The average age of admission was just a few weeks, so that would have been extremely unlikely.

Skuse: I think what we are therefore looking at is some environmental deficit that occurred at a period when such experience would be ubiquitous in the general population. It was this deficit which perturbed the development of a neural circuit that has eventually righted itself, insofar as the autistic features are ameliorating in later childhood. What was that experience? Following on from what I was suggesting yesterday about the importance of eye contact in social development, I wonder whether any of these infants had any significant face–face interaction with their ‘care givers’ during that early period. Infants have, as a matter of course, intense interest in eye-to-eye contact during the postnatal period.

Rutter: We know that they didn’t have much interaction, as shown by many reports from people visiting the institutions. The children were fed by bottles being stuck in their mouths and left there, and there were no toys. There was just one staff member for 30–40 children. One can be fairly sure that there was little opportunity for face–face interaction.

Skuse: We propose that there is a very primitive neural circuit that is anticipating such eye contact occurring, and this arouses huge interest in the infant within hours after birth. If this eye contact doesn’t happen for some sustained period of time, then there will be perturbations in this circuit. Some autistic features could well be consequential to that. The importance of this early face to face contact for neurodevelopment is pointed up by the LeGrand et al (2001) study of infants with congenital cataracts, who had impaired face processing abilities in later childhood.

Lord: It wouldn’t necessitate as much eye contact as middle-class children get in western societies. There are such cultural differences in how much time babies spend, for example, being carried on backs.

Skuse: I don’t know of any cultures in which mothers habitually carry their infants on their backs without making eye contact with their infants during feeding, for example.

Sigman: I have looked at this in African children because of some anthropological literature that suggested there is very little face–face eye contact.
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In fact, if you count the amount of eye contact that African babies have with their siblings, then their experience of eye contact is equivalent to that of American babies. A paper describing these results is in press in the *Journal of Cross-Cultural Psychology*.

*Bailey:* The Romanian adoptees are a very interesting sample. One assumes that the vast majority of babies went into the institution with normal brains, and that what followed then dramatically altered the course of development. What has struck me in hearing about them is the almost total environmental deprivation they were subjected to. They were just left alone for the vast majority of the day. I have no trouble in accepting that various experience-dependent mechanisms cause deficits in social interaction and communication. What I find particularly interesting is the presence of circumscribed interests. This comes to the point that Frankie Happé was raising earlier: there might be multiple routes to get to circumscribed interests. In autism it may be because particular brain systems are affected. It looks as if environmental deprivation—and its knock-on effects on social skills and communication—is another means for reaching circumscribed interests. It is interesting that the behaviour was there and that it has disappeared as the environment has become enriched.

*Rutter:* There are two caveats. First, we don’t know about prenatal alcohol exposure. This is likely to have been a problem in some. The other is the possible effects of severe stress on the mothers during pregnancy. I agree, though: in general we can assume that most of them will have had normal brains.

*Howlin:* Did the Romanian babies have eye contact with each other?

*Rutter:* They were mostly in separate cribs.

*Rogers:* As the children become ambulatory, leave their cribs and start to interact, can they begin to become social partners for each other in the way that siblings are in large families? Can the peers themselves begin to take the roles of other important people?

*Rutter:* That may well happen post-adoption, but I doubt whether that will have occurred pre-adoption because they were so delayed in development. Very few of them were even walking. The degree of deprivation in this group was profound.

References
