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LETTERS TO THE EDITOR

Aphasia, deafness, or mental retardation

Wilson *et al*[1] reported a new type of X linked mental retardation with 'striking aphasia' and other anomalies. I cannot find 'aphasia' indexed in three major databases.[2–4] In contrast, mental retardation occurs in 666 syndromes and deafness in 292.[4] A priori, therefore, the probability of language retardation being the result of aphasia rather than these two commoner conditions is remote. In order to establish a precedent for the use of 'aphasia' as a titular keyword, or when postulating a speech gene, it is all the more important to ensure that there is not the slightest hint of mental retardation or deafness. This was certainly not so in the cases reported.

I suspect that developmental aphasia is a rare variant of particular types of deafness. I suggested that the term 'developmental aphasia' be dropped unless peripheral ear disorders, including otitis media, can be excluded.[5] It is therefore ironic that all three cases had frequent respiratory infections, two having chronic or recurrent otitis. As for their hearing, it was not mentioned in case 1, and was said to be 'normal', at least in adolescence, in cases 2 and 3. Such cryptic information is virtually useless. To show the absence of a peripheral hearing defect a basic minimum protocol includes: (1) consistently normal pure tone audiometry, especially at high tones; (2) normal tympanometry and acoustic reflexes; and (3) no evidence that the above tests were abnormal earlier in life. This would certainly not have been true for the cases with otitis.

Even if these three criteria were fulfilled, it is still possible that unusual peripheral defects (for example, retrocochlear deafness) could be missed. It may not, of course, be easy or convenient to test such children. Nevertheless, no conclusions about rare or esoteric causes of speech or language defects can be drawn until any straightforward peripheral auditory dysfunction has been excluded.

Case 2 was said to be 'autistic'. Although autism has been associated with various syndromes (for example, rubella), most, if not all, of these syndromes also cause deafness,[6] which may in turn cause the autism.[7] Hence, like aphasia, there may be no justification for including autism in the title of another syndrome.

Another requirement for the diagnosis of aphasia is that the speech and language retardation is far below the general intellectual level, especially non-verbal IQ. Cases 2 and 3 were stated to have IQs of below 30 and 40, with no mention of which tests were used, or even if verbal or non-verbal. Verbally loaded tests like the Stanford-Binet are worse than useless since a specific verbal IQ deficit is confounded with overall low IQ. Case 1 at 3 years was said to have a developmental level of about 16 months with a vocabulary of five to 10 words. This sounds as if a standard developmental test was given, but there were no further details. To show aphasia, the language scale needs to be much lower than the other scales, otherwise aphasia and mental retardation are again confounded. Another X linked disorder was originally described as 'mental retardation-aphasia-shuffling gait-adducted thumbs, but aphasia was later reclassified as speech delay[4] or abnormality.[2] This is not surprising given that the index case[8] actually had higher verbal than non-verbal IQ (Stanford-Binet IQ 55, Raven IQ 41); hearing was not tested ('hearing appears to be grossly intact').

In view of general ignorance over the origin of language delay it is all the more important to distinguish the three rival causes, mental retardation, aphasia, and deafness. If clinical data, no matter how carefully collected, are reported in a muddled way that confounds these three causes, then readers may conclude that these distinctions are irrelevant.

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This letter was shown to Professor Wilson, who replies as follows.

Dr Gordon's argument that deafness is the explanation of 'aphasia' in our family with X linked mental retardation (XLMR)[1] is difficult to refute. Although all three of our affected males had 'normal' audiometry testing at the time of their initial evaluations for developmental delay, the actual audiograms and their timing regarding the history of chronic otitis media in two boys were not available for our review. It seems likely that significant hearing defects would have been noted by the parents or school/institutional personnel, but it is certainly true that sophisticated evaluation of hearing is worthwhile in those XLMR disorders where abnormal speech has been noted.

We used the term 'aphasia' in our title to emphasise our clinical impression that there was a dissociation between the degree of speech problems and cognition.[2–6] This was particularly evident in the older male who had a large sign language vocabulary—perhaps 'expressive aphasia' would have been a better term. I disagree strongly with Dr Gordon's opinion that mental retardation should be accepted as the cause of speech delay based on the use of keywords in databases. A long and current battle in the US concerns separation of mental retardation into specific causal entities, many with distinctive behavioural and neuropsychiatric phenotypes. One particularly instructive example is Williams syndrome in which chronic otitis media, hyperacusis, and a dissociation between language and cognitive function have all been noted.[3,4] Such disorders will guide us to the genes that account for male predominance and familial aggregation in language impairment.[5,6]

Progress in the delineation of XLMR has been remarkable over the past decade and it would seem negligent not to mention abnormal speech when it is striking to the clinical observer. Many of these observations may not hold up, as suggested by Paul *et al*[2] when they performed language assessments of fragile X syndrome adults in a blind fashion with controls having comparable degrees of mental retardation. Although the numbers of patients were small, their lack of discrimination contrasted with many clinical reports of specific language abnormalities in fragile X syndrome. The speech abnormalities mentioned in six other XLMR disorders[1] may also prove non-specific, but are worth pursuing in view of the open road to gene characterisation. Supporting Dr Gordon's view that abnormal speech in XLMR reflects either deafness or mental retardation is the conservation of human X chromosomes when compared to those of non-human primates with limited speech capacity. On the other hand, unusual evolutionary variation of a gene responsible for XLMR and abnormal speech might help explain our remarkable linguistic facility.

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Sex differences in the location of a spina bifida lesion

Three studies have shown that in patients with spina bifida the ratio of males to females is greater if the lesion includes only the lumbar or sacral region than if it includes the thoracic, cervical, or occipital spine.[1–4] A fourth study appeared to confirm this[5] but on further analysis[6] this was not found to be the case (J G Hall, personal communication). We sought to clarify the effect using data from Oxford.

Two series were studied: (1) derived from a survey of spina bifida births in 1965 to 1972

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