

**case report****Autism in Down's syndrome: family history correlates****M. Ghaziuddin***Taubman Centre, University of Michigan Medical Centre, Ann Arbor, Michigan, USA***Abstract**

Although the association of autism with Down's syndrome is said to be uncommon, several reports have described the co-occurrence of the two disorders. This report describes three additional cases of Down's syndrome with autism. In all the patients, a history suggestive of the broader phenotype of autism was obtained in parents. This suggests that familial factors specific to autism may play an important role even when autism complicates a known medical condition such as Down's syndrome.

**Keywords** Down's syndrome, autism, mental retardation

**Introduction**

Down's syndrome (DS) is a common genetic cause of mental retardation with a prevalence rate of 1-1.5 per 1000 live births. Ninety-five per cent of cases are caused by trisomy 21 and 5% are the result of translocation. Research on the behavioural aspects of DS has focused on three main areas: cognition, psychiatric comorbidity and personality (Flint & Yule 1994). People with DS are said to perform worse on verbal subtests of intelligence and better on visuospatial tests than mentally retarded controls

(see Flint & Yule 1994). A wide variety of psychiatric disorders have also been reported. Case-series and epidemiologic surveys have described the occurrence of several psychiatric disorders in DS such as attention deficit disorder (Green *et al.* 1989), depression (Warren *et al.* 1989) and schizophrenia (Lund 1988). Except for Alzheimer's disease, no neuropsychiatric condition has been specifically linked with DS. So far as temperament and personality are concerned, people with DS have been traditionally described as friendly, affectionate and extroverted. For example, Gibbs & Thorpe (1983) compared 49 non-institutionalized subjects with DS with 49 mentally retarded controls using a personality checklist. They concluded that 'the classic personality stereotype of the Down's syndrome child as affectionate and outgoing emerged', that the stereotype was unaffected by the race or sex of the child, and that the results provided further evidence in support of the concept of a DS personality. On the other hand, there is evidence that not all patients with DS possess the same personality characteristics. Clinical experience suggests that, while most people with DS possess a stable and compliant temperament, some 10-20% of patients show behavioural problems (see Flint & Yule 1994; Gibson 1978).

Consistent with the image of behavioural and social adaptability that people with DS often project, association of DS with autism is said to be

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described above, other mechanisms may also contribute to the association of autism with DS. For example, complicating physical conditions that occur in DS may contribute to the emergence of autistic features. Trisomy 21 causes a wide range of deficits and some of these may contribute to autistic symptoms (Howlin *et al.* 1995). Also, as we have discussed elsewhere (Ghaziuddin *et al.* 1992), certain pathophysiologic aberrations are common to both. For example, both autism and DS are associated with neuropathological findings such as heteropias or focal abnormal collections of grey matter in the cerebellar and cerebral cortices (Zellweger, 1977). These structural abnormalities, although not specific to either of the disorders, might reflect altered patterns of functional organization common to both.

In conclusion, although the association is said to be uncommon, autism can occur in people with DS. One reason why some people with DS develop autism may lie in the genetic vulnerability of these patients to autism, as reflected by an increased family history of autistic traits. These findings have relevance to the study of factors responsible for the association of autism with other medical conditions and raise the possibility that when autism co-exists with known medical disorders, autism-specific genetic factors may be important. Systematic family studies of the presence of autism with these disorders may clarify the contribution of familial/genetic factors to its aetiology.

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