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Cerebellar defect reported by Fragile X researchers

ARRI reported in Spring 1988 on magnetic resonance imaging (MRI) studies by Eric Courchesne and colleagues which revealed that two lobes of the cerebellum were abnormally small in autistic individuals. The October 27 issue of the *New England Journal of Medicine* presented two new studies which also used MRI to search for cerebellar defects linked to autism.

Allan Reiss reported in the *NEJM* that he has found a cerebellar defect in the brains of individuals with Fragile X syndrome which closely resembles the defect seen by Courchesne in autistic subjects. Fragile X, the second most common genetic cause of mental retardation, generally affects males and may cause more than 10% of all cases of autism.

Reiss studied four men with Fragile X syndrome and four normal controls. Scans done on Fragile X subjects showed "a statistically significant decrease in the size of the posterior portion of the vermis (lobules VI through X) as compared to the anterior portion (lobules I through V) . . . [in addition] the area of the pons is significantly smaller and the fourth ventricle significantly larger in the patients than in the controls." Other brain areas they studied appeared normal.

Reiss et al. compared their data with Courchesne's, and found that the defects seen in lobules VI and VII were remarkably similar in the two studies. In Courchesne's study, the average area of lobules VI and VII in autistic subjects was 249.4 square millimeters, while the average for controls was 304.9 square mm. In Reiss' study, the average for Fragile X subjects was 227.5 square mm, while the average for controls was 336.0 square mm.

Reiss says the two studies suggest "that the cerebellar vermis, particularly the posterior portion, is a component of the neurobiologic system that may be injured during critical periods of development in persons in whom autistic behavior develops." However, he noted that only two of his Fragile X subjects met the diagnostic criteria for pervasive developmental disorder, a disorder milder than, but similar to, autism.

Edward Ritvo et al. reported in the same issue of the *New England Journal of Medicine* that they had performed MRI scans on 15 autistic subjects (as well as 15 non-disabled controls), and had failed to find the defect reported by Courchesne. They speculate that their inability to replicate Courchesne's results may have been due to differences in MRI techniques, control groups, or the ages of the subjects.

In a reply to Ritvo's letter in the *NEJM*, Courchesne et al. suggested that the imaging techniques used in Ritvo's study may have "contributed to inaccurate measurement" and obscured any cerebellar defects. They reiterated that "when MRI data are acquired and analyzed according to our method, a subpopulation of adults with autism will be found to have a measurable decrease in the size of specific vermal lobules."

Asperger's syndrome: Is it autism?

by Alison Blake

He makes inappropriate, often quite irrelevant, remarks in company and appears gauche and childish.

He enjoys his job and his hobby, but is very sad and anxious because he is aware of his own social ineptness and would like to have friends and to marry.

He appears clumsy and ill-coordinated, has problems with buttons and laces, and is afraid of climbing.

He speaks in a pedantic style, in an accent quite unlike that of his local environment. For example, he referred to a hole in his sock as a "temporary loss of knitting."

The people described above by Lorna Wing in *Psychological Medicine* have a disorder called Asperger's Syndrome. While Hans Asperger first described this syndrome in 1944, researchers still debate whether Asperger's syndrome is a distinct disorder, or a mild form of autism—the high end of an "autistic spectrum."

The typical person with Asperger's is a "loner" who never quite fits in because of eccentric behavior, peculiar ways of speaking, and a lack of social skills. He or she is interested in social relationships, but suffers from what Wing describes as "a lack of ability to understand and use the rules governing social behavior."

People with Asperger's syndrome may graduate from regular schools and hold down jobs, but they are handicapped by their odd behavior and an autistic-like resistance to change. Ida Sue Baron studied seven children with Asperger's syndrome and found that "they were unable to form lasting friendships and children refused to return to their homes to play with them." She found that the older children "had over time withdrawn from the uncomfortable interactions which characterized their early years and retreated into the safety of their family or even isolated themselves from their family . . . . They felt rejected but did not understand how their behavioral responses contributed to their eventual position as social outcasts."

Unlike typically autistic children,

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Court rules autism not 'mental illness'

A U.S. District Court in California has ruled that insurance companies must consider autism as a developmental disability rather than a "mental illness," and provide coverage accordingly.

The decision arose from a suit brought by the parents of a child treated for 30 days at the Neuropsychiatric Institute at UCLA in Los Angeles. The Benefit Trust Life Insurance Company paid only $10,000 of the $54,696 bill for the treatment, claiming that the policy limited medical benefits for "mental illness or nervous disorders."

In ruling that autism is more appropriately categorized as "a disease or disorder of the brain," Judge Irving Hill noted that "medical research has produced increasingly strong evidence of a demonstrable organic basis for the syndrome" and that "there is a consensus among experts that . . . autism is not caused by environmental trauma."

ARRI thanks Dr. Edward Ritvo, who treated the child involved in this case, for informing us about this ruling.