Biomedical update:

Moebius, autism: link may indicate brainstem disorder

More evidence linking autism and Moebius syndrome is reported by Swedish researchers Christopher Gillberg and S. Steffenburg.

The researchers earlier published a case study (see ARRI 2/1) about a child with both autism and Moebius syndrome, a rare disorder of the skull nerves controlling the facial muscles. Moebius syndrome causes paralysis of both sides of the face and often results in vision difficulties, nervous system defects, speech problems, and a variety of arm and leg disorders.

Now Gillberg and Steffenburg report that of 17 patients with Moebius syndrome they have studied, 40 percent show all or many of the symptoms of autism. Only one-third of the individuals with Moebius syndrome were free from major behavioral problems.

"The association with autism and autistic symptoms is strong," the researchers say, noting that less than one percent of children in a typical population are autistic, compared to nearly half of the Moebius syndrome patients. Their findings, they say, "could be suggestive of a common underlying neurobiological deficit at the brainstem level."

It may be of interest, the researchers say, that none of the four females with Moebius syndrome in this study had characteristics of autism.

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"Autistic behavior in Moebius syndrome," C. Gillberg and S. Steffenburg, *Acta Paediatr. Scand.*, 78, 1989, pp. 314-316. Address: Christopher Gillberg, Barn- och ungdompsykiatriska kliniken, Box 7284, S-40235 Goteborg, Sweden.

Unusual CT findings reported

CT scans of the brains of nine healthy, high-functioning autistic men showed several possible defects, according to R. Jacobson et al.

Compared to scans of controls, scans of autistic subjects in this study showed enlargement of the third ventricle (one of four fluid-filled chambers in the brain). The researchers say this may indicate abnormalities in nearby brain pathways of brain "messenger" chemicals serotonin and noradrenalin; abnormal levels of both of these brain substances have been linked to autism.

Radiodensity studies showed lower densities in the left and right caudate nuclei in autistic individuals than in controls. The caudate nuclei, C-shaped structures lying deep within the brain, play a role in movement and possibly in cognitive processes.

The researchers say that if defects in these areas do exist, they are "selective," since no abnormalities were seen in the lateral ventricles or in the thalamus, which is near the caudate nuclei.

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"Selective subcortical abnormalities in autism," R. Jacobson, A. Le Couteur, P. Howlin and M. Rutter; *Psychological Medicine*, 18, 1988, 39-48. Address: R. Jacobson, Department of Psychiatry, Jenner Wing (G2) Level 1, St. George's Hospital Medical School, Cranmer Terrace, London SW17 0RE.

Rett's: muscle biopsies show abnormalities

Researcher Ahmad S. Teebi of Kuwait reports that muscle biopsies performed on three girls with Rett syndrome show changes in the mitochondria, small threadlike structures within cells that are the primary sources of cell energy.

The mitochondria "appeared abnormally swollen and dumbbell shaped," Teebi reports. "The membrane of some showed foamy vaculations, which may indicate membranous damage arising from an abnormality in the mitochondrial DNA."

The cause of Rett syndrome, a disorder which may affect only girls, remains a mystery. Scientists have found few significant chemical or structural abnormalities in the brains of children with this disorder, which causes autistic-like symptoms, severe growth and motor problems, loss of speech and mobility, chronic hand-washing or hand-wringing motions, and other symptoms.

Teebi says his findings may indicate that Rett syndrome is caused by an X-borne gene mutation which causes the death of males before development; females may survive because they carry a normal gene on their second X chromosome. On the other hand, adverse interactions between a normal and abnormal X chromosome in girls could cause the defect; in this case boys would be unharmed since they would not have two interacting X-chromosome genes.

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"Rett syndrome: genetic clues based on mitochondrial changes in muscle," Ahmad S. Teebi, *American Journal of Medical Genetics*, 32, 1989, pp. 142-144. Address: Ahmad S. Teebi, c/o Sadika A. Al-Awadi, Director, Kuwait Medical Genetics Centre, P.O. Box 4080, Safat, 13041, Kuwait.

Many schizophrenic children show early autistic symptoms

The debate over the relationship (if any) between autism and schizophrenia continues, with a new study by John Watkins et al. indicating that "a high proportion of schizophrenic children have early symptoms that meet . . . criteria for infantile autism."

The authors emphasize that while a surprisingly large number of schizophrenic children in their study were autistic, only a very small percentage of autistic children later become schizophrenic.

When Watkins and his colleagues studied the early histories of 18 consecutive patients with childhood schizophrenia, they found that 39% of the children had symptoms of autism before developing schizophrenia. In these cases, autistic symptoms continued after schizophrenia developed.

The researchers also found that the schizophrenic children with a history of autistic symptoms had an earlier onset of schizophrenia than the children with no history of autism. In fact, no cases of schizophrenia occurred before the age of nine except in children with previous symptoms of either autism or "childhood onset pervasive developmental disorder" (a diagnosis given to children exhibiting relatively mild autistic symptoms which begin after three years of age).

Noting that both autism and schizophrenia are associated with a wide range of causes ranging from infections to genetic defects, the authors speculate that "some risk factors might . . . contribute to the development of both disorders."

The authors also report that severe language problems and motor problems such as poor muscle tone and lack of coordination were seen in 72% of the children before symptoms of schizophrenia appeared. Symptoms of depression frequently appeared around the time that schizophrenia was diagnosed.

"Our results indicate," the researchers conclude, "that the early histories of schizophrenic children are far from benign . . . . Overall, there were far more severe symptoms and far more social impairment prior to the onset of schizophrenia in these children than is seen in the childhood histories of schizophrenic adults."

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"Symptom development in childhood onset schizophrenia," John M. Watkins, Robert F. Asarnow, and Peter E. Tanguay; *Journal of Child Psychology and Psychiatry*, Vol. 29, No. 6, 1988, pp. 865-878. Address: John M. Watkins, Division of Child Development and Biobehavioral Sciences, Department of Pediatrics, University of California, Los Angeles, CA 90024.