Rett Syndrome: diagnostic guidelines offered

A major achievement in recent years has been the subdividing of the population labelled "autistic" into smaller subgroups, such as Fragile X autism, lyase deficiency autism, purine autism and Rett Syndrome. These subgroups have different causes and, presumably, will be found to have different treatments.

To assist physicians in distinguishing one of these subgroups, Rett Syndrome, from undifferentiated autism, Andreas Rett and Bo Olsson have developed a set of diagnostic guidelines.

Rett and Olsson found that symptoms present in Rett Syndrome but NOT in the general autistic population included:

—slow movements and hypoactivity;

—stereotypic hand movements including wetting hands with saliva, making "washing" motions, continually bringing hands together, and flexing and stretching the middle finger joints;

—episodes of hyperventilation, only through the mouth;

—no proper chewing; and,

—"sensory-motor intelligence" level equivalent to that of a four- to six-month-old normal child.

Behaviors seen only in autism and NOT in Rett Syndrome include:

—a wide variety of movements, including swiftly alternating movements, and proper gripping, gait and chewing;

—ability to say at least three words or one clause, or to use echolalia;

—stereotypic behaviors such as manipulating objects, clapping, tapping or flapping hands, rocking, tip-toeing, self-stimulating behaviors involving the face, and self-injury; and,

—"sensory-motor intelligence" at least equal to that of an 11- to 14-month-old.

Rett and Olsson believe the differences between symptoms of Rett Syndrome and symptoms of autism indicate that children with Rett Syndrome have overall severe retardation involving the higher cortical functions, while autism is more likely to be caused by abnormalities in the lower brain.

Sarojini S. Budden, of the Oregon Health Sciences University, notes that symptoms common to Rett Syndrome but not to autism include:

—willingness to make eye contact;

—inability to use the hands to manipulate objects or pick up food;

—development of appropriate speech before the onset of symptoms;

—severe problems with chewing and swallowing food; and,

—early onset and frequent occurrence of seizures.

He stresses the importance of diagnosing Rett Syndrome early, noting that "for many families, the diagnosis has been crucial in understanding their children's gradual deterioration in spite of their continued efforts in providing their children with appropriate care."

Of special interest was the report by Budden that one of his 13 Rett Syndrome patients had a Fragile X (p22) abnormality.

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"Behavioral Observations Concerning Differential Diagnosis Between the Rett Syndrome and Autism," Bo Olsson and Andreas Rett; *Brain and Development*, Vol. 7, No. 3, 1985. Address: Andreas Rett, Neurologisches Krankenhaus der Stadt Wien-Rosenhugel-Abteilung fur entwicklungsgestorte Kinder, 1130, Wien, Riedelgasse 5, Austria.

—and—

"Rett Syndrome: Studies of 13 Affected Girls," Sarojini S. Budden; *American Journal of Medical Genetics*, 24:99-109, 1986. Address: Sarojini S. Budden, Crippled Children's Division, P.O. Box 574, Portland, Oregon 97207.

Syndrome linked to metabolic defect

An international research team reports that Rett Syndrome may be caused by a defect resulting in a deficiency of biotin, one of the B vitamins (Bachmann et al.).

Tests of the urine of one Rett Syndrome patient indicated abnormal levels of several enzymes called carboxylases. The researchers gave the patient biotin supplements for one month, and were able to correct the abnormal carboxylase levels. (Because the patient died suddenly of unrelated causes, the research team was not able to report long-term results of the therapy.)

The researchers suggest that "an early supplementation of biotin to patients with Rett Syndrome should be considered, especially if indications of disturbed carboxylase activity are found."

Richard Haas and fellow researchers at the University of California at San Diego found that six of seven Rett Syndrome patients they studied had elevated levels of blood pyruvate, indicating a defect in carbohydrate metabolism. When these patients were placed on a ketogenic (high fat, low carbohydrate) diet, five had fewer seizures, five showed behavior and motor improvement, and six gained weight.

Editor's note: If the relationship between increased biotin need and Rett Syndrome is confirmed in other patients, we would have three B vitamins related to autism. The relationships between folic acid and Fragile X, and between vitamin B6 and undifferentiated autism (30% to 40% are helped), have been well established in multiple studies.—B.R.

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"Rett Syndrome Revisited: A Patient with Biotin Dependency," C. Bachmann, J.P. Colombo, J. Schaub, B. J. Burri, L. Sweetman, and B. Wolf; *European Journal of Pediatrics*, 144:563-566, 1986. Address: C. Bachmann, CZL, Inselspital, CH-3010, Bern, Switzerland.

—and—

"Therapeutic Effects of a Ketogenic Diet in Rett Syndrome," Richard H. Haas, Marylynne A. Rice, Doris A. Trauner and T. Allen Merritt; *Am. Journal of Med. Genetics*, 24:225-246, 1986. Address: Richard H. Haas, UCSD Med. Ctr. (H-815-B), Ped.Neur., 225 Dickinson St., San Diego, California 92103.