Biomedical update:

Peptide studies show three autism subtypes

Different patterns of peptides (chains of amino acids which combine to form proteins) in the urine of autistic children "clearly indicate three subtypes of the autistic syndrome," according to Norwegian researchers (Reichelt et al.) who analyzed urine samples from 145 autistic subjects.

One group of test subjects, whose members developed autistic symptoms following periods of normal development, had peptide levels similar to those found in people with slow-onset schizophrenia. Another group of children with "classical" autism had peptide levels resembling those of children with attention disorders and hyperactivity.

Many of the subjects' parents reported that their children's autistic symptoms became worse after ingestion of cow's milk or grain products. This may indicate that the children's bodies were not producing the enzymes necessary to break down the peptides formed from these foods.

A separate study (Israngkun et al.) of 14 autistic children found that levels of many peptides were abnormally high. The researchers believe one particular peptide, which appeared in elevated levels in all autistic subjects, may prove to be a biological marker for autism.

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"Childhood Autism: A Complex Disorder," K.L. Reichelt, G. Saelid, T. Lindback, and J.B. Boler, *Biological Psychiatry*, 21:1279-1290, 1986. Address: K. L. Reichelt, Pediatric Research Institute, Rikshospitalet, 0027 Oslo 1, Norway.

—and—

"Potential Biochemical Markers for Infantile Autism," Porn Israngkun, Howard Newman, Suman Patel, Valentine Duruibe and Hussein Abou-Issa, *Neurochemical Pathology*, Vol. 5, 1986. Address: Howard Newman, Div. of Clinical Chemistry, Dept. of Pathology, The Ohio State University, Columbus, Ohio 43210.

Chromosome defects found

While several abnormalities of the X and Y chromosomes have been found in autistic individuals, few abnormalities of autosomal (non-sex linked) chromosomes have been described. However, U.S. researchers (Mariner et al.) recently reported finding four cases of autism with different autosomal chromosome abnormalities.

The patients in the report were both autistic and retarded, and had minor face and body abnormalities such as "funnel chests", high arched palates, prominent ears, and extra digits. Their autistic behaviors included marked speech delays, echolalia, resistance to change, fascination with inanimate objects, self-stimulatory behavior, tactile defensiveness, and avoidance of eye contact.

Abnormalities on chromosomes 3, 5, 16 and 17 were found by the researchers.

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"Autism, Mental Retardation, and Chromosomal Abnormalities," Robin Mariner, Andrew Levitas, Randi J. Hagerman, Ann C. M. Smith, Alfred W. Jackson, III, Rebecca Berry, and Marcia Braden; *Journal of Autism and Dev.Disorders*, Vol. 16, No. 4, pp. 425-440, 1986. Address: Randi Hagerman, The Child Dev. Unit, The Children's Hospital, 1056 East 19th Avenue, Denver, Colorado 80218.

First diagnosis: most parents want more information

Most parents of children with severe developmental delays are dissatisfied with the manner in which their children were first diagnosed, according to a 1986 English study (Quine and Pahl). The researchers concluded that:

1. Parents want to be told as early as possible that there is cause for concern, even if an exact diagnosis cannot be made. Parents told after a long delay, and particularly after a period of seeming evasion by their physicians, were the most dissatisfied.
2. Parents are experts about their own children, and "their observations and information are of paramount importance in reaching a diagnosis."
3. Families value a sympathetic and caring approach by doctors and other medical personnel.
4. Parents like to be told about their children's disabilities "in private, together, and with the child present if possible."
5. Parents want more information about their children's disorders. Seventy-four percent of the parents surveyed said they had not been given enough information by their doctors. Because parents often are too shocked at the time of the initial diagnosis to ask questions or remember all of what is said, follow-up visits should be scheduled with the physician or another professional who is sympathetic and knowledgeable about appropriate services.

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"First Diagnosis of Severe Mental Handicap: Characteristics of Unsatisfactory Encounters Between Doctors and Parents," Lyn Quine and Jan Pahl; *Soc. Sci. Med.*, Vol. 22, No. 1, pp. 53-62, 1986. Address for either author: University of Kent at Canterbury, Health Services Research Unit, Canterbury, Kent, England.

Tuberous sclerosis link debated

Tuberous sclerosis—a hereditary disease causing tumors on the surfaces of ventricles in the brain, hardened areas on the brain surface, mental deterioration and epileptic attacks—has been reported from time to time to occur in conjunction with autism.

However, Mark Greenstein and Suzanne Cassidy at the University of Connecticut have found only one documented case in which tuberous sclerosis and autism apparently occurred in the same individual. They add that while children with tuberous sclerosis may show some autistic behaviors, "the confusion of symptoms of tuberous sclerosis with those of autism may occur for a brief period only, and these conditions can be distinguished on clinical grounds alone.

"Considering the lack of supportive reports corroborating a link between tuberous sclerosis and autism, we question its continued inclusion in the differential diagnosis of autism," they concluded.

Wayne Fisher and fellow researchers in North Dakota disagree, saying that a recent survey involving only residents of their state identified three adults and one child with tuberous sclerosis who also met criteria for "pervasive developmental disorder", a category which includes autism and milder but similar disorders. If co-occurrence of the two disorders were just chance, they say, it should only happen in six cases per billion people.

Editor's note: Of the more than 9,000 autistic children whose case records are on file at the Institute for Child Behavior Research, there are at least a dozen with tuberous sclerosis. The association arises much too often to be regarded as a coincidence, in my opinion. When resources are available, the relationship will be investigated in depth.—B.R.

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"Is Tuberous Sclerosis a Cause of Autism?" (letter), Mark A. Greenstein and Suzanne B. Cassidy; *New England Journal of Medicine*, 314:449, February 13, 1986. Address for either author: University of Connecticut Health Center, School of Medicine, Farmington, Connecticut 06032-9984.

—and—

"Tuberous Sclerosis and Autism" (letter), Wayne Fisher, Paul Kolstoe, Jacob Kerbeshian, and Larry Burd; *Developmental Medicine and Child Neurology*, 1986, 28, 814-823. Address: Wayne Fisher, Grafton State School, Grafton, North Dakota 58237.