Biomedical Update:

Fragile X: check for eye problems

A new study emphasizes the importance of eye exams for individuals with Fragile X syndrome, a genetic abnormality strongly linked to autism and retardation.

Dominick Maino and colleagues evaluated the ocular problems of 30 children and teenagers with Fragile X syndrome, and found that:

— 30% exhibited strabismus (inability of the eyes to align properly, due to muscle defects). Most of these were cases of esotropia, or inwardly crossed eyes.

—59% of the eyes evaluated exhibited a significant degree of hyperopia (farsightedness), 17% myopia (nearsightedness), and 22% astigmatism (irregular curvature of the cornea and lens of the eye, leading to faulty vision).

Maino notes that only two other studies on ocular problems in Fragile X have been published, and both showed a high incidence of strabismus in individuals with the syndrome. While the cause of eye defects in Fragile X is unknown, he says, "it has been suggested that the connective tissue dysplasia [abnormal development of the tissues that bind organs and other body parts] which occurs in this syndrome may result in the development of moderate to high refractive error and strabismus."

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"Optometric findings in the Fragile X Syndrome," Dominick M. Maino, Michael Wesson, Darrell Schlange, Gerhardt Cibis, and Joseph H. Maino; *Optometry and Vision Science*, Vol. 68, No. 8, 1991, pp. 634-640.

New report on treatment for hair-pulling

ARRI 5/2 reported on the successful use of Prozac (also see cover story in this issue) to treat trichotillomania, or compulsive hair-pulling, in an autistic teenager. A new case study by Mohammad Ghaziuddin and colleagues indicates the drug haloperidol, commonly known by the brand name Haldol, may be equally successful in treating this behavior problem.

Ghaziuddan's patient, an 11-year-old boy who was both autistic and severely retarded, had been pulling his hair out for about two years. The researchers report that "his head was covered with numerous bald patches covering at least 70% of the scalp." Behavior modification procedures and other drugs (Mellaril and Ritalin) were unsuccessful in controlling the behavior.

One week after starting Haldol, the researchers report, the boy stopped pulling his hair and appeared calmer. An attempt to reduce the dosage after six months (from 2 mg daily to 1.5 mg) was followed by a reappearance of the behavior, which stopped again when the higher dosage was reinstituted. The researchers have now followed the case for nine months, and report that "[the boy's] hair-pulling has completely disappeared, although his temper tantrums and occasional aggressive outbursts continue."

Ghaziuddan and colleagues conclude that "when trichotillomania occurs in autism and mental retardation, and when it is not accompanied by depression or obsessive-compulsive symptoms, haloperidol may be a useful medication to try."

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"Brief report: haloperidol treatment of trichotillomania in a boy with autism and mental retardation," Mohammad Ghaziuddin, Luke Y. Tsai, and N. Ghaziuddin; *Journal of Autism and Developmental Disorders*, Vol. 21, No. 3, 1991, pp. 365-371. Address: M. Ghaziuddin, CAPH, Box 0390, Univ. of Michigan Hospitals, 1500 E. Medical Center Dr., Ann Arbor, MI 48109-0390.

Swedish report: An autism subgroup?

Swedish researchers Christopher Gillberg et al. say tests have revealed a specific chromosome abnormality in six boys with a combination of autism, retardation, and mild to moderate physical defects.

The researchers say the boys had IQs below 55, avoided eye contact, and (in all but one case) had poor muscle tone. All of the boys fulfilled the criteria for autism, with the exception of one profoundly retarded boy who had significant autistic behaviors. Unusual physical features seen in some or all of the subjects included large protruding ears, high-arched palate, short stature, low weight, kyphoscoliosis (curvature of the spine from back to front and from side to side), and epilepsy. While symptoms varied, Gillberg says, "the overall picture was such that one senses the presence of a true 'chromosomal syndrome.'"

The chromosome defect seen in all six boys is a partial trisomy of chromosome 15; instead of having the normal two copies of chromosome 15, the boys had an additional copy of a portion of the chromosome. "These findings," Gillberg and colleagues say, "further underline the necessity to screen for chromosomal abnormalities in all cases of autistic syndromes with mental retardation without a known cause."

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"Autism associated with marker chromosome," Christopher Gillberg, Suzanne Steffenburg, Jan Wahlstrom, I. Carina Gillberg, Agneta Sjostedt, Tommy Martinsson, Sigrun Liedgren, and Orvar Eeg-Olofsson; *Journal of the American Academy of Child and Adolescent Psychiatry*, 30:3, May 1991, pp. 489-494. Address: Christopher Gillberg, Child Neuropsychiatry Centre, Box 17113, S-402 61 Goteborg, Sweden.

Buspirine, high-serotonin diet tested for aggression

A combination of buspirone (an anti-anxiety medication that alters the brain's uptake of serotonin and other substances) and a special diet significantly decreased a retarded adult woman's aggression, according to a recent report by A. Gedye of Canada.

Gedye administered 10 to 20 mg per day of buspirone to the 39-year-old woman, discontinued the drug, and then reinstated it (eventually increasing the dosage to 25 mg) along with a special diet high in serotonin and low in high-protein foods. Chocolate, caffeine, and high-salt foods were removed altogether.

During the drug-only phase, Gedye reports, the woman's aggression decreased by 67%; when the drug was discontinued, her tantrums increased sharply. Treatment with buspirone plus diet control caused "a further reduction of 85% compared with baseline," Gedye says, and "when episodes with self-injurious movements were added [to the data analysis], the same pattern of reduction emerged." While increased aggression and self-injury appear to be correlated with menstrual cycles in many developmentally disabled women, no changes in the woman's behavior during her menstrual cycle were seen either on or off the drug.

George Realmuto et al. tested buspirone on four autistic children in 1989 (see ARRI 4/3), and found that the hyperactivity of two children decreased, and one exhibited fewer stereotypical behaviors. The fourth child did not improve.

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"Buspirone alone or with serotonergic diet reduced aggression in a developmentally disabled adult," A. Gedye, ***Biological Psychiatry***, 30, 1991,. pp. 88-91. Address: A. Gedye, Ministry of Social Services and Housing, 9 E. Columbia St., New Westminster, BC, V3L 3V5, Canada.

Researcher Jaak Panksepp of Bowling Green University has established a foundaton which will provide funds for research into new treatments for autism. The Lost Children's Fund, established in memory of his daughter, will also be involved in other aspects of brain research, as well as in research into social and emotional problems affecting children. Donations to the BGSU Lost Children's Fund can be mailed to the Mileti Center, Bowling Green State University, Bowling Green, OH 43403.