Biomedical/Psychology Update:

British journal editorial recommends widespread Fragile X screening

Noting that fragile X syndrome is the second leading cause of identifiable mental retardation, an editorial by D. W. York Moore in the *British Medical Journal* calls for widespread screening of "at risk" populations including individuals with severe learning disabilities and those in special education programs.

The researcher says that "fragile X is not diagnosed as commonly as might be expected" given its prevalence, and says "this may reflect the fact that the syndrome has attracted little publicity: few doctors and even fewer members of the public seem to know anything about it." He notes that the recent development of an accurate test for fragile X makes such testing practical.

Fragile X, a constriction on the X chromosome, has been tentatively linked to autism and learning disorders in addition to retardation. Often the syndrome is diagnosed only after a family has several affected children. While the disorder primarily affects males, recent research indicates that some girls with fragile X show cognitive deficits and exhibit behavioral problems such as odd communication, social isolation, and peculiar mannerisms.

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"New developments in the fragile X syndrome: time to consider screening?," D.W. York Moore, *British Medical Journal*, July 25, 1992, Vol. 305, No. 6847, p. 208.

Editor's note: It may prove to be important to identify fragile X cases if preliminary findings are borne out. We are aware of about 10 cases of fragile X who showed significant improvement on high dose vitamin B6 and magnesium. If you have information on similar cases, please contact ARI.

Autism, Down's: is association common?

While the co-occurrence of autism and Down's symdrome is generally believed to be rare, M. Ghaziuddin and colleagues propose that "the association is perhaps more common than generally believed."

The researchers report seeing three individuals with both Down's and autism at their clinic in one year. Two were from the same school district, which serves an area with about 40 children with Down's; this suggests, Ghaziuddin et al. say, that "for every 100 persons with Down's syndrome, there may be about four or five with coexisting autism."

Their findings, they say, are consistent with a study by Christopher Gillberg and colleagues which found that 5% of children with Down's syndrome had coexisting autism, and a study by J. Lund which found that 10% of 44 adults with Down's syndrome also had autism.

"Although no causal relationship can be proposed between the two conditions," they say, "it is interesting that certain pathophysiologic aberrations are common to both. For example, both autism and Down's syndrome are associated with neuropathological findings such as...focal abnormal collections of gray matter in cerebellar and cerebral cortices. These structural abnormalities, although not specific to either of the disorders, might reflect the altered patterns of functional organization common to both." And it is possible, the researchers say, that trisomy 21—the chromosome defect that causes Down's syndrome—may also be linked to autism.

"It is important that the diagnosis of autism is not missed when it occurs in Down's syndrome," Ghaziuddin et al. say, because children with both disorders may fare best when placed in structured programs capable of dealing with autistic behaviors.

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"Autism in Down's syndrome: presentation and diagnosis," M. Ghaziuddin, L.Y. Tsai, and N. Ghaziuddin; *Journal of Intellectual Disability Research*, 1992, 36, 449-456. Address: M. Ghaziuddin, Child and Adolescent Psychiatry-Taubman Center, Box 0390, University of Michigan Medical Center, 1500 E. Medical Center, Ann Arbor, MI 48109-0390.

ABRs recommended

Auditory brainstem response tests (ABRs) are recordings, from electrodes placed on the scalp, of brainwaves generated in response to auditory stimuli such as clicks. While autism researchers use the test to explore possible brainstem defects, Ami Klin of Yale says they may be overlooking another phenomenon revealed by their studies: the presence of peripheral hearing loss in a significant number of autistic subjects.

Klin reviewed 11 ABR studies, and found that "56 to 78 of the 170 subjects identified as autistic showed suggestive evidence of hearing abnormalities," even though studies often excluded subjects with known hearing problems. Klin says that other evidence—for instance, the high rate of ear infections in autistic children, and non-ABR studies showing hearing problems in many autistic subjects—indicates that autistic individuals may have an unusually high rate of hearing defects.

Noting that standard hearing tests are imprecise and often difficult to perform on autistic children, Klin says that "the indications of peripheral hearing abnormalities revealed in the ABR studies...argue very forcibly for the use of the ABR technique in the assessment of the hearing status in children with autism, particularly in those cases where autism is associated with mental retardation or neurological disorder."

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"Auditory brainstem responses in autism: brainstem dysfunction or peripheral hearing loss?," Ami Klin; *Journal of Autism and Developmental Disorders*, Vol. 23, No. 1, March 1993. Address: Ami Klin, Yale Child Study Center, Yale University School of Medicine, P.O. Box 3333, New Haven, CT 06510.

What's in a look? Eye contact and autism

British researchers say that autistic children do not use eye contact normally to determine another person's goals, and that this may be a clue as to why they have so much difficulty understanding the mental states of other people.

Wendy Phillips and colleagues tested normal infants, autistic children ages 3-5, and mentally handicapped children in two "ambiguous" situations (in which the experimenter's goal was not clear to the child) and one "unambiguous" situation. The three test situations were:

—a "blocking" task, in which the experimenter waited until the child was playing with a toy, and then covered the child's hands to prevent further playing.

—a "teasing" task, in which the experimenter offered the child a toy, but withdrew it when the child reached for it. After withholding the toy for a few seconds, the experimenter returned it to the child.

—a "giving" task, in which the experimenter simply gave the child a toy.

"All of our normal infants, from 9 to 18 months, responded to the teasing and blocking task by instantly establishing eye contact with the experimenter," the researchers say. "So did the majority of children with mental handicap...In contrast, most children with autism, with an equivalent mental age, did not make eye contact as often with the experimenter during the critical periods. Instead, they looked at the experimenter's hand, or at the toy, and in the blocking test simply tried to remove the hand obstructing their own."

Phillips et al. believe that non-disabled children use eye contact in ambiguous situations as "a primitive psychology with which to make sense of past actions and predict a person;s next action." They note that all of the normal subjects made eye contact immediately following the tease or block tests, but less than half of them did so following the unambiguous "giving" test.

The researchers cite research showing that autistic children show normal eye contact when requesting something, and do not avoid eye contact in general. Rather, they say, "they appear not to use it for the same purposes as nonautistic children."

Phillips et al. say that autistic children's apparent failure to use eye contact to diagnose a person's goal may reflect an abnormality in an early stage in the development of a "theory of mind"—that is, the ability to understand that other people know, want, feel or believe things, and to predict others' behavior based on such knowledge.

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"The role of eye contact in goal detection: evidence from normal infants and children with autism or mental handicap," Wendy Phillips, Simon Baron-Cohen, and Michael Rutter; *Development and Psychopathology*, 1992, 4, pp. 375-383. Address: Wendy Phillips, Department of Child and Adolescent Psychiatry, Institute of Psychiatry, University of London, London SE5 8AF, England.