Autism, Mental Retardation, and Chromosomal Abnormalities

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There are reports of sex chromosomal abnormalities including XXY, XYY, and fragile X karyotypes in autistic individuals, but structural autosomal defects have rarely been reported. This paper presents four patients with autism, mental retardation, minor dysmorphic features, and structural autosomal defects. These patients shared autistic features including fascination with inanimate objects, catastrophic reactions to changes in their environment or their daily routine, echolalia, and poor relatedness; IQ scores indicate mild to severe retardation. Their autosomal abnormalities included inversion/duplications of 3p and 16q, 5p+, and 17p−. Parental chromosomes were all normal. Chromosomal analysis should be performed on mentally retarded, autistic individuals, especially those with minor physical anomalies and no specific etiology for their retardation.

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INTRODUCTION

The first description of autism cited features including an inability to relate to people and situations in an ordinary way, excellent rote memory, obsessive desire for sameness, normal cognitive potential, and intelligent but aloof parents (Kanner, 1943). Because symptoms were often present from early infancy, Kanner postulated an "inborn defect." The current definition of autism recognizes pervasive perceptual, language, and behavioral problems, in addition to difficulties in relating to other people and objects. Seventy-five percent of autistic children can be expected to perform at a retarded level throughout life (Havelkova, 1968; Rutter, 1970). Cold parents are no longer implicated in the etiology of autism; instead, a variety of biological problems have been associated with the widespread CNS dysfunction of autism. Autism occurs in up to 7.4% of individuals with congenital rubella (Chess, 1977), and in association with infantile spasms (Riikonen & Amnell, 1981), cerebral lipoidosis (Creak, 1963), and phenylketonuria (Ciarnello, VandenBerg, & Anders, 1982; Knobloch & Pasamanick, 1975).

Autism has also been associated with various sex chromosomal abnormalities. In 1971 Abrams presented a case history of an autistic male with an XYY karyotype (Abrams & Pergament, 1971). Crandall reported several XXY patients with many features of autism (Crandall, Carrol, & Sparkes, 1972). Judd found an abnormally long Y chromosome in 3 of 8 autistic patients (Judd & Mandell, 1968); Hoshino found a similarly long Y in 9 of 22 autistic males (Hoshino et al., 1979). The fragile X syndrome has the highest incidence of autism of any documented syndrome or chromosomal abnormality. Autism has been reported in 69% of 23 fragile X males studied (Levitas, McBogg, & Hagerman, 1983) and in 16% of a larger population of fragile X males (Hagerman, Jackson, Levitas, Rimland, & Braden, 1986).

The relationship of autosomal abnormalities with infantile autism has been less well characterized. Siva-Sankar reported that chromosomal breaks were found 3.5 times more frequently in autistic patients than in a group composed of schizophrenics, psychotics, and patients with primary behavior disorders (Siva-Sankar, 1970). Hansen reported a case history of an autistic female with a translocation between chromosome #22 and a chromosome in the D group (Hansen, Brask, Nielsen, Rasmussen, & Sillesen, 1977). In addition to these reports of structural chromosome abnormalities in association with autism, this disorder has been documented in individuals with trisomy 21 (Wakabayashi, 1979) and trisomy 22 (Turner & Jennings, 1961; Bieselee, Schmid, & Laulis, 1962).

Presented here are four autistic, mentally retarded patients with various structural autosomal abnormalities, including 3p +, 5p +, 16q +, and 17p -.