Preliminary Observations on Language and Learning in XXY Boys*

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This study investigated the relationships that exist between the sex chromosome aneuploidies in males (ie, XXY, XYY) and certain aspects of development. A group of infants with these chromosome lesions and a control group were located through a chromosome screening of male neonates [1, 2). These children were followed in a long-term study with the purpose of identifying reactive styles or developmental lags that might be related to the sex chromosome variations. The data, obtained through semistructured parental interviews (recorded verbatim) and direct observations of the child, were reduced to 15 operationally defined categories of behavior that represented the child's development and functioning in the most relevant aspects of psychosocial adaptation [3]. Preliminary conclusions based on the systematic sampling method were tested independently by formal psychologic evaluations and developmental assessments.

This report presents data pertinent to the speech/language development, cognitive performance, and early academic learning of 13 XXY and 9 control children. The methods employed in this study and the development of these children during the first 3 years of life were reported previously [3].

Infants with chromosome variations were located through a chromosome screening of newborn males. In all cases the parents were informed about the chromosome abnormality and the known developmental implications. Prospective parents were informed prior to the screening procedure that they would be notified if their infant was found to have a chromosome abnormality.

Parents of infants with the XXY karyotype were told that the phenotypic consequences of this chromosome variation would probably be testicular

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changes and delayed puberty. The limited knowledge about the behavioral implications of these karyotypic variations was explained also. Emphasis was placed on the need for long-term prospective studies of children with these variations in order to determine whether affected individuals carried any risk for developmental impairment.

The need to share this information with the parents determined the nature of the control sample, which was comprised of *neonates* having a chromosome abnormality that did not affect the phenotype adversely (ie infants with familial balanced autosomal rearrangements). These infants also were included since their parents had to be informed about the existence of a variation. It was possible to give the parents considerable reassurance about the insignificance of the chromosome variation for the child's development, by indicating that one of the parents carried the same variation and that it had not affected their development adversely. Thus, the information given the families about the chromosome variation and its possible developmental significance was different for the aneuploid and the control groups; however, this was the most adequate control possible for the variable of parental informing. The control population was matched with the aneuploid group for socioeconomic status and race.

EARLY SPEECH AND LANGUAGE DEVELOPMENT

By 36 months, 9 of the 13 children with the XXY chromosome complement were reported by parents to manifest a delay in speech and language development; reports of delays were seen in only 1 of the 9 controls ($p \leq .05$; Wilcoxon Rank-Sum Test). For the 9 XXY children, the parents reported delays in sentence-building skills, difficulties with speech sound production (articulation), problems with intonation and accent, or difficulties in finding specific words with which to express thoughts clearly. The parents of one control child reported difficulties with speech sound production.

Formal speech and language assessment on the XXY children revealed that receptive (understanding) language skills were age appropriate. However, reductions were present in immediate auditory memory skills. In addition to reduction in the amount of information that the children could retain at any one time, these children manifested deficits in preserving the serial order of this auditorily presented material.

The XXY boys evidenced various problems in expressive language skills. Deficits in syntax were evident. The children demonstrated word-finding problems (dysnomias), especially under confrontation-naming requests. The children also evidenced problems in creating a coherent, organized, and developed narrative; in all instances, however, the content of the narrative was appropriate.

The specific aspects of communication disorders in the XXY boys were confirmed recently in a study by Graham and his colleagues [4]. Graham et al compared the communication skills of a group of 14 unselected XXY boys (13 from this study; 1 from another study) with a group of 15 control boys matched for race, age, educational level, and socioeconomic status. No differences between groups were apparent on screening tests of hearing sensitivity. However, differences in certain auditory-processing abilities were noted. The XXY group demonstrated significant decrements in auditory memory. Auditory-rate-processing abilities were compromised in 64% of the XXY boys. The XXY group had more difficulty than the controls with phonemic discrimination of similar words in the presence of background noise.

Receptive language abilities were age appropriate for the XXY group, except for comprehension of certain syntactic structures, which was below expectation for age. Expressive language abilities were relatively more impaired than receptive language abilities for the XXY group. Developmental histories revealed that the XXY boys had significantly more difficulty remembering the names of commonly used words and in telling a story. Of particular interest, then, was the observation that these word-finding difficulties persisted into later ages. The XXY children demonstrated significant deficits in the production of syntax. Speech sound production skills did not differ between the groups.

COGNITIVE DEVELOPMENT

On the Bayley Scales of Mental Development, administered at 20-24 months of age, the Mental Scale scores did not distinguish between XXY and control groups. However, expressive language skills were significantly behind comprehension in 10 of 13 XXY children; a similar discrepancy was seen in only 2 of the 9 controls ($p = .0159$; Fisher exact probability test). In contrast to the reduced Verbal scores, XXY children performed consistently above chronologic age level expectations on various form board tasks.

The Wechsler Scales of intelligence were administered to the 13 XXY and 9 controls at 5-7 years of age. Prior to age 6 the Wechsler Preschool and Primary Scale of Intelligence was used; after age 6 the WISC-R was employed.

There was no significant difference between the means of the Full Scale IQ or of the Performance IQ for the XXY and controls ($p > .10$; *t* test). However, there was a significant difference in the Verbal IQ means ($p < .01$), with the XXY group demonstrating a lower mean score than the controls (Table 1).

A comparison of the Verbal and Performance scores for the 13 XXY children revealed a discrepancy in the direction previously noted with the Bayley Scales (Table 2). In all but one instance (Child 11, Table 2), the Full Scale Wechsler scores were in the average to superior range. However, there was a

TABLE 1. Mean IQ Scores

	N	Full Scale IQ	Verbal IQ	Performance IQ
XXY	13	110.20	101.46*	117.50
Controls	9	114.77	113.00	113.55

* p. .0175 (t test).

TABLE 2. Disparity Between Verbal and Performance Scale Scores on (he WISC/WPPSI for XXY Children

Child	Age	Full Scale IQ	Verbal Scale IQ	Performance Scale IQ	Disparity	Sig. Lev*
1	7:4	115	105	122	+17	<.05
2	7:4	99	89	111	+22	<.01
3	6:2	93	86	101	+15	<.01
4	7:7	128	121	129	+8	
5	5:6	115	101	127	+26	<.001
6	6:3	111	100	120	+20	<.001
7	6:4	111	104	118	+ 14	<.01
8	5:o	128	116	135	+ 19	<.01
9	5:7	118	102	131	+29	<.oOl
10	7:o	101	97	106	+9	
11	6:8	76	79	79	0	
12	6:5	105	94	117	+23	<.001
13	6:2	133	125	132	+7	

*Sattler J: "Assessment of Children's Intelligence." New York: Holt, Rinehart&Winston, 1975, P 450, Table C-18. This table gives the probability of obtaining the designated differences between the individual Verbal and Performance IQs on the WISC and WPPSI.

significant discrepancy between the Verbal and Performance Scale scores in 9 of the 13 boys (69% of the XXY children), with the Performance Scale scores being consistently higher than the Verbal Scale scores. One boy (Child 11) was in the borderline range of intelligence in the Verbal, Performance, and Full Scale scores.

The Full Scale IQ scores of the 9 control children were in the average to superior range (Table 3). However, only 2 boys (Children 3 and 7, Table 3) showed a significant disparity between the Verbal and Performance Scale scores. In one boy (Child 7) the Performance score was higher than the Verbal; in the second boy (Child 3) the Verbal score was higher than the Performance.

Twelve of the 13 XXY children are beyond the first grade at the time of this report; one child is presently in that grade. Eleven of the 12 XXY children beyond grade one were referred independently by the teachers for learning disability evaluations because of difficulties in learning to read and to spell. Nine

TABLE 3. Disparity Between Verbal and Performance Scale Scores on the WISC/WPPSI for Controls

Child	Age	Full Scale IQ	Verbal Scale IQ	Performance Scale IQ	Disparity	Sig. Lev
1	6:6	111	108	112	+4	
2	5:8	108	110	104	- 6	
3	6:8	100	108	90	- 18	<.01
4	5:10	134	130	131	+1	
5	5:4	116	117	112	- 5	
6	7:1	120	120	115	- 5	
7	6:11	122	107	133	+26	<.001
8	5:6	115	114	114	0	
9	5:8	107	103	111	+8	

of the 11 were enrolled subsequently in special education programs for children with learning disabilities. The remaining 2 are receiving private tutorial assistance in reading. Only 2 of the 13 XXY children are doing well in their early school learning tasks. One of the 2 is presently in the first grade and appears to be acquiring early reading decoding and arithmetic skills.

Eight of the 9 control children are beyond the first grade at the time of this report; one child is presently in grade one. Only one control child (who is presently in the second grade) has demonstrated a significant learning disability and is presently receiving private tutorial assistance.

Eight of the XXY children who manifested learning disabilities in school also had problems with early speech and language development; they also showed a significant discrepancy between the Verbal and Performance IQ scores (with lower Verbal scores) in the 5–7-year-old IQ testing (Table 4). Three of the XXY boys with learning disabilities were not consistent with respect to the speech/language or IQ discrepancy variables. Two of the XXY children without learning disabilities also did not manifest early speech and language problems or demonstrate a Verbal/Performance Scale discrepancy on the 5–7-year-old Wechsler Scales.

Six of the control children who are doing well in their early learning skills also manifested no previous speech or language problems and demonstrated no Verbal/Performance Scale discrepancy in the 5–7-year-old IQ testing (Table 4). One control child who showed no speech/language difficulties, but did manifest a Verbal/Performance Scale discrepancy (with a *higher* Verbal Scale score), has progressed well with his early learning. The only control child with a significant early learning disability also showed a Verbal/Performance Scale discrepancy (with a higher Performance Scale score) but did not manifest an earlier speech or language problem. The one control child who showed a speech problem (ie a pronunciation difficulty) has done well academically and did not demonstrate a Verbal/Performance Scale discrepancy.

TABLE 4. Relationship Between Early Speech/Language Development, Performance on 5–7-Year-Old IQ Testing, and Early Academic Learning

	Problems With Early Speech/Language Development		Verbal/Performance IQ Discrepancy in 5–7-year-old IQ Testing		Early Learning Disability	
	Present	Absent	Present	Absent	Present	Absent
XXY	8	1	a		a	
		1			1	
	1					
		2		2		2
Controls		6		6		6
		1	1*			1
			1			

*Verbal Scale score significantly higher than Performance Scale score.

DISCUSSION

A number of reports concerning XXY children and adults ascertained from biased and unbiased samples have noted that the affected individuals suffered frequently from developmental speech and language delays [5–15]. Furthermore, when the Verbal and Performance IQ values were compared, it was found frequently that XXY individuals showed Performance IQ scores that were higher than Verbal IQ [6, 11, 13–17], although this was not a consistent finding [5, 18–20]. These results are the reverse of those found in girls with Turner syndrome (ie sex chromosome monosomy; 45,X), where the Verbal IQ scores tend to be higher than the Performance IQ scores [14, 18, 19, 21–24].

The relationship between language disorders and reading-spelling difficulties has been well established. The association is clearly evident in our XXY group. Although one control child with a reading disability did not demonstrate an early speech or language delay, he did show a discrepancy in the Verbal and Performance IQ values (with a lower Verbal IQ score).

The results are far too limited to justify any long-range conclusions or predictions about future development. However, the preliminary data, when considered in relation to cross-sectional observations on older Turner syndrome patients, raise the possibility that pathologic variations in sex chromosome complements (XXY, XO) may be associated with specific developmental changes in the central nervous system.

These observation must be subjected to future studies in order to devise remedial techniques for early intervention with XXY children, before language and reading difficulties become chronic.

SUMMARY

Difficulties with processing, storing and retrieving, and producing linguistic information were particularly evident among the XXY boys. These decrements in abilities were associated with decreased achievement in a variety of reading and spelling tasks in this study as well as in that of Graham et al [4]. The data suggest that a continuum of language disability is present throughout the early life of these children and is associated with the XXY anomaly. Furthermore, when considered in relation to cross-sectional observations on older Turner syndrome patients, these data suggest the possibility that pathologic variations in sex chromosome complements may differentially affect brain development and subsequent functioning of the children.

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