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Fragile X syndrome: speech and language characteristics

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FRAGILE X SYNDROME: SPEECH AND LANGUAGE CHARACTERISTICS

by

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Presented in partial fulfillment of the requirements for the degree of Master of Arts

UNIVERSITY OF MONTANA

1984

Approved by:

[Signature]
Chairman, Board of Examiners

[Signature]
Dean, Graduate School

August 13, 1984
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Chapter I

LITERATURE REVIEW

Fragile X syndrome is a chromosomal disorder responsible for a significant proportion of male mental retardation. Diagnosis of this disorder has become more common in the past fifteen years. Cytogenetic study is the only means of diagnosis at the present time. Reports of the clinical manifestations of fragile X syndrome vary considerably. Some investigators report that the speech and language characteristics of affected men and boys are strikingly similar. Others have observed communication behaviors much like those of the mentally retarded population as a whole. The importance of communication evaluation for diagnosis and effective referral has not been established. The presence or absence of a characteristic set of speech and language symptoms will affect the role the speech pathologist plays in referral, diagnosis, counseling, and treatment for clients and their families.

Fragile X Syndrome

Fragile X syndrome is one of at least four X-linked conditions resulting in male mental retardation (Turner and Opitz, 1980; Gerald, 1981). It involves an anomaly, or fragile site, on an X chromosome. A fragile site is an area of weakness at a specific point on a chromosome.
that may become a break (Hecht, Hecht and Glover, 1981). Most fragile sites are not associated with physical abnormalities (Hecht, 1979). Fragile X chromosome does appear to be associated with physical abnormalities and mental retardation.

The disordered chromosome, called Marker X, is identified by the constriction, or fragile site, on the long arm (Howard-Peebles, Stoddard and Mims, 1979). This narrowing is at a specific region, at band 27 or 28, near the end of the long arm (Martin, Mathies and Lowry, 1980). Breaks may result in satellites seen in cytogenetic study (Hecht, Glover and Kaiser-Hecht, 1982). The fragile X condition is designated fra(X) (q27) or fra(X) (q28), noting the fragile site, X chromosome, long arm, and band number.

Fragile sites are identified by karyotyping 50 to 100 cells obtained from blood samples. Fragile site expression is not complete. That is, fragile sites may not be observed in 100% of cells studied (Herbst, 1980). The proportion of fragile X expression is dependent upon the culture media in which cells are grown for study. Expression is best for culture media deficient in folic acid and thymidine (Hecht, 1979).

Cytogenetic study of mentally retarded males has revealed 4% to 50% fragile X expression (Howard-Peebles, Stoddard and Mims, 1979; Jacobs, Glover, Mayer, Fox, Gerrard, Dunn and Herbst, 1980). Random chromosome deviation in normal patients may occur, and 4% expression is the pres-
ently accepted criteria for diagnosis of fragile X syndrome (Lahr, 1984). Female carriers may not show fragile X expression, particularly after age 30 (Jacobs et al., 1980). Proportion of fragile X expression does not appear to be correlated with level of intelligence in males, although it is strongly correlated with mental ability in carrier females (Chudley, Knoll, Gerrard, Shepel, McGahey and Anderson, 1983).

Fragile X syndrome is a frequent cause of male mental retardation, and it occurs at a rate of .92/1000 (Brown, Friedman, Jenkins, Brooks, Wisniewski, Ragnuthu and French, 1982). It may be second only to Down's syndrome, which occurs at a rate of 1.4/1000, as an identifiable cause of male mental retardation (Lahr, 1984).

**X-Linked Mental Retardation**

Several factors suggest that one or more X-linked disorders are responsible for many cases of male mental retardation. Frequently, the pattern of inheritance is such that the disorder occurs only in males, yet is transmitted to the child by the mother, a carrier (Valentine, 1975). The carrier does not demonstrate clinical disease. This pattern suggests that the gene associated with the disorder is located on an X chromosome, passed from mother to son. The disorder is expressed in the male child because a normal gene is not present to counteract the disordered gene. The chance of recurrence of the X-linked condition is 25% for each birth.
It has been estimated that there are 10% to 50% more males than females in the mentally retarded population (Gerald, 1981; Hecht et al., 1981). Mentally retarded males are more likely to have mentally retarded male relatives than mentally retarded females are to have mentally retarded female relatives (Gerald, 1981).

Many male mentally retarded persons exhibit no major physical abnormalities, metabolic disease, or central nervous system disorder (Herbst, 1980; Gerald, 1981). Until recently, no chromosomal disorder associated with these cases was evident. The term nonspecific X-linked mental retardation is used to describe male patients demonstrating X-linked patterns of inheritance, relatively minor physical aberrations, and no known etiology. Persons affected with fragile X syndrome were among those in this category before diagnostic methods allowed identification of the defective X chromosome.

Hecht, Hecht and Glover (1981) estimated that nonspecific X-linked mental retardation occurs approximately once in 600 live births. Fragile X chromosome may account for approximately one-half of X-linked mental retardation (Lahr, 1984).

Clinical Symptoms

Mental Retardation

Borderline to profound mental retardation of fragile X patients has
been reported. McDermott, Walters, Howell and Gardner (1983) observed intelligence quotients (IQ) in the 15 to 43 point range. Subjects in a study by Chudley et al. (1983) demonstrated IQs in a range from 10 to 66. Lahr (1984) reported that most fragile X patients' mental abilities are in the moderately retarded range, although Chudley et al. reported most in the severely to profoundly retarded range.

Two males with fragile X syndrome demonstrating normal mental abilities were described by Daker (1983). Hecht and Jacky (1983) indicated that fragile X males with normal mental abilities may exist, but stated that such cases had not been documented sufficiently. Carrier females have demonstrated normal to borderline mental retardation (Hecht et al., 1981).

Physical Characteristics

The physical features of fragile X syndrome are subtle and variably expressed (McDermott et al., 1983). They may not be present in all affected members of the same family (Herbst, 1980). There appear to be no major physical anomalies associated with X-linked mental retardation, and minor anomalies have not been consistently found listed in the medical files of affected males (Herbst, 1980). Herbst (1980) reported that minor physical features may not be unusual enough to elicit comments in medical files.
Macro-orchidism

One frequently reported symptom of fragile X syndrome is macro-orchidism, or large testicular volume. Volumes two to six times that of normal males have been reported (Brown et al., 1982).

Facial and Body Characteristics

Jacobs et al. (1980) observed that the facial appearance of fragile X patients tends to be "cast in the same mold", and is characterized by large ears and a prominent mandible. They noted that these features are seen in normal men as well, and are not solely indicative of fragile X retardation.

McDermott et al. (1983) listed many physical features in case descriptions of patients with fragile X syndrome. These included facial asymmetry, high forehead, broad nose, hypoplastic maxilla, prominent mandible, high palate, large simple ears, epicanthal folds, dysmorphic hands and fingers, scoliosis, neck webbing, gynecomastia, cafe' au lait spots, and heterochromia of the irises. Large head circumference, long narrow face, large "lop" ears, and high birth weight have also been reported (Levitas et al., 1983; Herbst, Dunn, Dill, Kalousek and Krywan-iuk, 1981; Turner and Opitz, 1980).
Speech and Language

Speech and language delays have been documented in studies of males with fragile X syndrome. However, disagreement exists as to whether a verbal disability greater than that predicted by cognitive ability is typical of fragile X syndrome. Some investigators reported that a characteristic cluster of speech behaviors is associated with the disorder.

Turner and Opitz (1980) stated that Lehrke "was strongly impressed by a verbal deficit" among patients with X-linked mental retardation. Howard-Peebles et al. (1979) observed a generalized language disability in fragile X patients indicated by performance scores which averaged 12.8 points better than verbal scores on the Weschler Adult Intelligence Scale. These patients demonstrated strengths on the Illinois Test of Psycholinguistic Abilities in nonverbal skill areas.

Herbst et al. (1981) found that individuals with fragile X syndrome demonstrated poorer receptive vocabulary skills and nonverbal reasoning than men with X-linked mental retardation not expressing fragile X. They did not observe a general verbal disability in their subjects.

Herbst (1980) reviewed cases of fragile X syndrome reported in four previous studies. An overall verbal expression deficit in excess of that expected based on borderline to profound levels of mental retardation was not evident. No test results other than cytogenetic findings
were consistent enough for diagnosis of fragile X syndrome.

An unusual pattern of speech has been observed in males with fragile X syndrome. In a study of seven Canadian families, which included twenty-seven adults and seven boys, Jacobs et al. (1980) reported that "there seems to be little doubt that the fragile X chromosome is associated with a clinically recognizable syndrome consisting of a variable degree of mental retardation, a characteristic repetitive, jocular speech, macro-orchidism, and the fragile site on the distal part of Xq." These authors found patients' speech so characteristic that "one retarded boy, encountered in another context, and one adult, encountered in the course of our studies, were thought to have this form of MR on the basis of their speech." Such reports suggest that speech and language observations may be significant diagnostic factors for fragile X syndrome.

Turner, Daniel and Frost (1980) reported that fragile X patients tend to have a particular quality about their speech which they described as narrative and compulsive "litany" speech. A more detailed description of this speech characteristic was not presented in the report.

McDermott et al. (1983) provided speech and language descriptions of twelve adult members of seven families. The subjects, affected with fragile X syndrome, had IQs in a range of 15 to 35, and were aged 23 to 68 years. Some descriptions of patients' communicative characteristics
were similar to those described by Jacobs et al. The following are descriptions of the communicative behaviors of several individuals. 1) single words, short phrases, good articulation, some echolalia 2) disjointed, syllabic, perseverative speech 3) continuous production of simple repetitive sentences with defective articulation; distinctly unusual, rapid and repetitive speech; jargon and coprolalia 4) constant, repetitive, perseverative speech. The investigators reported that no set of speech characteristics was typical of their subject sample. In fact, one subject with vocabulary ability at the eight year level, demonstrated "normal" speech and articulation.

Newell, Sanborn and Hagerman (1983) used a battery of tests (i.e., Sequenced Inventory of Communication Development, Peabody Picture Vocabulary Test - Revised, Test of Language Development, Goldman-Fristoe Test of Articulation, Apraxia Battery, language sample) to assess the speech and language functioning of twenty-one subjects with fragile X syndrome aged 17 months to 31 years. The subjects demonstrated syntactic ability consistent with mental age, common articulation errors, dysfluency, and poor auditory memory. They also demonstrated perseverative and echolalic speech, and incomplete sentence production, with short "outbursts" of two to three word phrases.

Levitas, Hagerman, Braden, Rimland, McBogg and Matus (1983) investigated that communicative behaviors of autistic children with fragile X syndrome. Most "striking" about the speech of these children was its observational character. The authors stated that this characteristic of
speech was seen in nonautistic fragile X syndrome children as well. Autistic fragile X syndrome children demonstrated typical "autistic features" in their speech which included echolalia, immature grammar, abnormal speech melody, and a high percentage of noncommunicative utterances (Levitas et al., 1983). Additional characteristics which the researchers had observed previously in nonautistic fragile X patients, and in the subjects as well, included dysfluencies, paraphasic substitutions, and a pattern of short, explosive bursts of observational comments.

The terms used to describe the speech characteristics of people with fragile X syndrome (e.g., litany, jocular) have not been well defined. In addition, reports of communicative behaviors of patients were often made by professionals other than speech pathologists. Physicians and others who have recorded this information may have differing criteria for evaluating the speech and language behaviors of their mentally retarded patients. Thus, a certain amount of ambiguity about the communicative functioning of fragile X patients exists.

Purpose

The purpose of this study is to describe the speech and language characteristics of some children diagnosed as having fragile X syndrome. The syndrome affects a number of people the speech pathologist is reasonably likely to serve in clinical or school settings. Knowledge of the disorder may lead to more effective assessment and intervention for
these clients. The literature included some disagreement as to whether fragile X patients demonstrate communicative behaviors different from those of other mentally retarded people. The present study will lend more information about this group of individuals. It is hoped that this study will promote

1) greater understanding of the disorder among speech pathologists, and

2) more detailed and controlled investigation of fragile X syndrome patients by professionals trained in speech pathology.

Specific information of interest in the present investigation included the following:

1) Levels of verbal development demonstrated relative to other areas of development.

2) Frequency of unusual speaking behaviors (e.g., repetitive and narrative speech) similar to those reported in the literature.

Oregon Health Sciences University, Child Development and Rehabilitation Center provided multidisciplinary assessment for several fragile X syndrome patients. The information from these assessments was examined and described relative to the literature. This study is based on descriptions of speech and language behaviors. Many were recorded by physicians. It represents a summary of the most salient communicative behaviors demonstrated by a small group of children with fragile X syndrome.
Chapter II

METHODS

Subjects

The subjects were seven male patients with a diagnosis of fragile X syndrome. The patients had been referred to the Child Development and Rehabilitation Center of the Oregon Health Sciences University for evaluation in Genetics Clinic between 1982 and 1983. Chronological age at the time of diagnosis ranged from 3 years, 5 months to 18 years, 8 months. The subjects resided in various locations throughout the state of Oregon. They had previously received multidisciplinary evaluation through Child Development Clinic, Cerebral Palsy Clinic or Multiple Discipline Evaluation Clinic which are evaluation programs in the Child Development and Rehabilitation Center. These evaluations occurred between 1969 and 1983, and included pediatric, speech and language pathology, special education, physical therapy, occupational therapy and nursing assessment.

Two additional subjects were diagnosed with fragile X syndrome during the same period of time at Oregon Health Sciences University. They were chronologically too young to demonstrate the characteristics of interest, and were excluded from this study.
Procedures

The files of patients diagnosed with fragile X syndrome were examined. Information in four areas was collected if it was available: (1) speech and language (2) psychology (3) motor (4) cytogenetic. Speech and language test results, and descriptions of communication by parents, speech pathologists and other professionals were examined.

The descriptions are of previously conducted evaluations. Therefore, detailed information about testing procedures, controls and results were not always available. Due to the small number of subjects, statistical analysis of the data was not performed. The purpose of the study is to describe the speech and language characteristics of these subjects with fragile X syndrome.
RESULTS

Language

No isolated verbal deficit was indicated by comparing levels of language ability with levels of cognitive and motor development of the subjects in the sample. Each of the seven subjects demonstrated delayed receptive language abilities. For these subjects, levels of receptive language ability were within 6 months of cognitive and motor skills measured at the same time.

Expressive language ability was consistent with receptive language ability in the seven subjects. Expressive language ability was measured at a level within 6 months of receptive language ability for all subjects. One pediatric report for J. S. suggested that his expressive language ability was significantly more delayed than his gross motor ability, however, language age levels were not provided for that evaluation. Receptive and expressive language levels, and level of mental retardation, are presented in Table 1.
Speech

Several speech characteristics which were suggested in the literature appeared in the files of subjects in this study. These characteristics included poor speech intelligibility, echolalia, perseverative speech, and inappropriate random talking. Frequency of the four speaking behaviors of interest are presented in Table 2.

Intelligibility

Poor speech intelligibility was reported for four of the seven subjects. The following are descriptions of poor speech intelligibility obtained from the files. Chronological ages are indicated in parentheses. Sources of the descriptions are indicated by underlining.

S.G.: (CA 9:0) speech pathologist: muttered unintelligibly; jargon still evident; when a question was posed to him he would respond and continue with his jargon.
(CA 16:6) physician: frequently hard to understand.

P.H.: (CA 5:0) physician: speech unclear but understandable, poorly articulated speech.
speech pathologist: distorted sibilants and blends
(CA 18:8) speech pathologist: several sound substitutions; imprecise, cluttered speech with some mumbling;
most verbalization intelligible and adequate for conversation.

S.B.: (CA 3:1) **physician**: slow speech with slurring.
(CA 6:0) **speech pathologist**: vocalized a great amount, most of it being unintelligible; tends to verbalize at an unusually rapid rate, running words together; speech is quite infantile.

J.W.: (CA 4:11) **speech pathologist**: unintelligible jargon; intelligible single words.

**Echolalia**

Echolalia was reported for four of the seven subjects. The following descriptions of echolalic behavior were obtained from the files. The term echolalia was used in describing each of the four subjects.

S.G.: (CA 5:3) **physician**: echoic, 3 year level speech; three to four word sentences.
**speech pathologist**: jargon and echolalia are still very much evident in this child's speech. During observation, the boy was heard to cue in repeatedly on the final word of questions put to himself and to his classmates. (For example, Linda, what color are your shoes? S: Shoes, shoes, shoes.) He echoed peer responses throughout the
observation period.

(CA 9:0) physician: There is still much mimicking and repeating of things.

(Echolalia was also observed when S.G. was evaluated at the ages of 11:2, 14:0 and 16:6.)

P.H.: (CA 6:4) speech pathologist: demonstrated echolalia when he reached the ceiling items on language tests.

S.B.: (CA 3:1) psychologist: Instead of responding to conversation he frequently repeats what was said.

(CA 6:0) speech pathologist: There is some echolalia present. For example, when he is given a sentence to repeat, he tends to echo only the last couple of words. Also, other statements that are made to him of a casual nature tend to be imitated.

J.W.: (CA 13:0) physician: J. is developmentally delayed, and is in special education placement. Some of his greatest disabilities are in the area of expressive language. He appeared quite echolalic today on examination.

Parent report regarding T.H. suggested echolalia, however, this behavior was not indicated in evaluation reports.

T.H.: (CA 4:3) parent: He doesn't talk because he doesn't
want to talk. He won't answer a question which is asked him, but he repeats it. He could talk if he wanted to because he can imitate.

Perseverative Speech

Two of the seven subjects were reported to repeat their own utterances inappropriately. The term perseverative was used specifically in each of these files. The following descriptions of perseverative speech were obtained from the files.

S.G.: (CA 5:3) speech pathologist: repeatedly echoed single words three to four times. 
(CA 9:0) physician: There was a lot of repetition of words and phrases.

nurse: He speaks with one or two words, often repeating them over and over again.

(CA 11:2) psychologist: Speech was further echoic and perseverative at times, though age appropriate on rare occasions.

P.H.: (CA 5:3) school administrator: He speaks in a somewhat gutteral manner, repeating his sentences two or three times as if he cannot stop.
Inappropriate Random Talking

Inappropriate random talking was noted in the files of two subjects.

S.G.: (CA 9:0) **physician**: Chatter and phrases; some of it made sense and some of it did not make so much sense. . .
His comment such as "going to day camp tomorrow" does bear some relationship to his mother having told him he is going to day camp. There were some phrases about J. and streakers and naked which had something to do with conversation recently in the home.

**nurse**: His phrases are seemingly unrelated to events occurring around him.

**Psychologist**: S. constantly talked to himself, saying things like "let's go home, let's go home".

**speech pathologist**: During the exam S. continuously verbalized inappropriately.

(CA 11:2) **physician**: Echoic and unrelated talking; his behavior during the day consists of aimless wandering and continual chattering to himself; talked randomly to himself.

P.H.: (CA 18:6) **physician**: When P. gets real tired he may talk to himself.

(CA 18:8) **speech pathologist**: Frequently rambled and
got off the main topic.

Cognitive Functioning

Cognitive abilities of the subjects were in the mildly to moderately retarded range as measured by psychologists. The range of intelligence quotients, available for five subjects, was 52 to 75. Results of cognitive evaluations are presented in Table 3.

Behavior was not consistent across subjects although several behaviors (e.g., distractibility) were common to some subjects. Behavioral observations included the following descriptions.

S.G.: Distractible, short attention span, stereotyped behaviors, purposeless activity, impulsivity.


S.B.: Perseveration of his own rather aimless activities, emotional outbursts, negativistic, destructive behaviors, hyperactive, temper tantrums, self-aggression, affable but slow in mental status, impulsive

J.W.: Highly distractible.

Subject S.G. was diagnosed as having a severe thought disorder at age 11 years, 2 months. His speech was described by the examining psychologist as "rapid, chronic, garbled, somewhat incomprehensible and bizarre in content". A high degree of variability of language expression, most of which was bizarre and meaningless in content, was reported.

Motor Ability

Motor delays were noted in the charts of six of the seven subjects. Information regarding the motor development of one subject, T.H., was unavailable. Levels of motor ability are presented in Table 4.

Cytogenetic Results

Cytogenetic evaluations revealed 9% to 50% expression of Marker X. Fragile X expression is presented in Table 5.

Physical Characteristics

The facial and body characteristics of the seven children in this study included several which had been described in the literature. None of these characteristics were seen in every subject in the sample. Physical characteristics included large lop or cupped ears, low set and
rotated ears, simple pinna pattern, large genitalia, macro-orchidism, macrocephaly, prominent forehead, mandible and nose, flat midface, hypertelorism, and epicanthal folds.
Table 1
Receptive and Expressive Language Levels

<table>
<thead>
<tr>
<th>Subject</th>
<th>Chronological Age (years:months)</th>
<th>Receptive Language</th>
<th>Expressive Language</th>
<th>Level of Retardation</th>
</tr>
</thead>
<tbody>
<tr>
<td>S.G.</td>
<td>5:3</td>
<td>2:0/2:6</td>
<td>2:0/2:6</td>
<td>moderate</td>
</tr>
<tr>
<td>S.B.</td>
<td>6:0</td>
<td>3:0/4:2</td>
<td>3:0</td>
<td>mild/mod.</td>
</tr>
<tr>
<td>J.B.</td>
<td>3:2</td>
<td>1:0/1:6</td>
<td>1:0/1:6</td>
<td>moderate</td>
</tr>
<tr>
<td>J.W.</td>
<td>3:11</td>
<td>1:5/1:9</td>
<td>1:6</td>
<td></td>
</tr>
<tr>
<td></td>
<td>4:1</td>
<td>1:8</td>
<td>1:6/1:8</td>
<td>moderate</td>
</tr>
<tr>
<td></td>
<td>4:2</td>
<td>2:0*</td>
<td>*</td>
<td></td>
</tr>
<tr>
<td>J.S.</td>
<td>0:7</td>
<td>0:5*</td>
<td>*</td>
<td></td>
</tr>
<tr>
<td></td>
<td>1:8</td>
<td>1:0</td>
<td>0:8</td>
<td>(IQ=57, no classif.)</td>
</tr>
<tr>
<td>T.H.</td>
<td>2:8</td>
<td>1:9/1:10</td>
<td>1:6/1:8</td>
<td>mild</td>
</tr>
<tr>
<td></td>
<td>4:3</td>
<td>2:0/2:2</td>
<td>1:6/1:8</td>
<td></td>
</tr>
</tbody>
</table>

*Developmental profile measure of communication did not separate receptive and expressive language levels.

Table 2
Frequency of Four Speaking Behaviors

<table>
<thead>
<tr>
<th>Subject</th>
<th>Poor Speech Intelligib.</th>
<th>Echolalia</th>
<th>Perseverative Speech</th>
<th>Random Talking</th>
</tr>
</thead>
<tbody>
<tr>
<td>S.G.</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>P.H.</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>S.B.</td>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
</tr>
<tr>
<td>J.B.</td>
<td>X</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>J.W.</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>J.S.</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>J.H.</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>4/7</td>
<td>4/7</td>
<td>2/7</td>
<td>2/7</td>
</tr>
</tbody>
</table>

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Table 3
Cognitive Abilities

<table>
<thead>
<tr>
<th>Subject</th>
<th>IQ</th>
<th>Mental Age/ Chron. Age</th>
<th>Test</th>
<th>Level of Functioning</th>
</tr>
</thead>
<tbody>
<tr>
<td>S.G.</td>
<td>75</td>
<td></td>
<td>PPVT</td>
<td>moderate/trainable</td>
</tr>
<tr>
<td>P.H.</td>
<td>60</td>
<td>4:0/8:3</td>
<td>Stanford/ Binet</td>
<td>mild/mod.</td>
</tr>
<tr>
<td>S.B.</td>
<td>53</td>
<td>3:0/6:0</td>
<td>Stanford/ Binet</td>
<td>mild/mod.</td>
</tr>
<tr>
<td>J.B.</td>
<td>--</td>
<td></td>
<td></td>
<td>moderate</td>
</tr>
<tr>
<td>J.W.</td>
<td>57</td>
<td>2:5/4:2</td>
<td>Stanford/ Binet</td>
<td>moderate</td>
</tr>
<tr>
<td>J.S.</td>
<td>54</td>
<td></td>
<td>Bayley Scales</td>
<td></td>
</tr>
<tr>
<td>T.H.</td>
<td>--</td>
<td></td>
<td></td>
<td>mild</td>
</tr>
</tbody>
</table>

Table 4
Motor Abilities

<table>
<thead>
<tr>
<th>Subject</th>
<th>Chronological Age (years:months)</th>
<th>Fine Motor</th>
<th>Gross Motor</th>
</tr>
</thead>
<tbody>
<tr>
<td>S.G.</td>
<td>5:5</td>
<td>1:9/2:0</td>
<td>2:6/3:0</td>
</tr>
<tr>
<td>P.H.</td>
<td>6:4</td>
<td>1:10/2:0</td>
<td>3:0/3:6</td>
</tr>
<tr>
<td>S.B.</td>
<td>3:1</td>
<td>2:6</td>
<td>2:0</td>
</tr>
<tr>
<td>J.B.</td>
<td>3:2</td>
<td>1:4/1:6*</td>
<td>*</td>
</tr>
<tr>
<td>J.W.</td>
<td>4:2</td>
<td>0:4</td>
<td>0:4</td>
</tr>
<tr>
<td>J.S.</td>
<td>0:7</td>
<td>0:11</td>
<td>0:11</td>
</tr>
<tr>
<td>1:0</td>
<td>1:8</td>
<td>1:1</td>
<td>0:11</td>
</tr>
<tr>
<td>2:11</td>
<td></td>
<td>2:6</td>
<td></td>
</tr>
<tr>
<td>T.H.</td>
<td>--</td>
<td>--</td>
<td>--</td>
</tr>
</tbody>
</table>

*Developmental profile measure did not separate fine and gross motor ability levels.*
Table 5

Cytogenetic Test Results

<table>
<thead>
<tr>
<th>Subject</th>
<th>Cells Expressing Per Cells Studied</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>S.G.</td>
<td>25/50</td>
<td>50</td>
</tr>
<tr>
<td>P.H.</td>
<td>13/50</td>
<td>26</td>
</tr>
<tr>
<td>S.B.</td>
<td>25/75</td>
<td>33</td>
</tr>
<tr>
<td>J.B.</td>
<td>15/75</td>
<td>20</td>
</tr>
<tr>
<td>J.W.</td>
<td>15/50</td>
<td>30</td>
</tr>
<tr>
<td>J.S.</td>
<td>13/52</td>
<td>25</td>
</tr>
<tr>
<td>T.H.</td>
<td>9/100</td>
<td>9</td>
</tr>
</tbody>
</table>
Chapter IV

DISCUSSION

One of the most notable communicative characteristics of the subjects in the present study is the similarity of language functioning and mental ability. Like subjects in the Newell, Sanborn and Hagerman (1983) study, these seven subjects demonstrated language levels commensurate with levels of mental retardation.

The subjects demonstrated four speech behaviors (i.e., reduced speech intelligibility, echolalia, perseverative speech, and inappropriate random talking) which are demonstrated by other groups of mentally retarded people. Darley (1978) stated that slowness in maturation is reflected in motor and cognitive skills when a generalized deficiency is present. The subjects in the present study demonstrated cognitive, motor and language disabilities from which reduced speech intelligibility could be predicted. Fay (1980) stated that a child having extensive problems in the area of language development may exhibit greater echoic output than is normal, in both the number of utterances and length of echoes. This behavior reflects difficulty with comprehension, greater number of increments in short term memory which accompanies physiological maturation, and increased exposure to verbal stimulation (Fay, 1980). Rosenburger (1978) stated that echolalia may
be most noticeable when other propositional speech is lacking.

Perseverative speech is also commonly observed in mentally retarded children. Karlin, Karlin and Gurren (1965) reported that mentally retarded children demonstrate perseverative speech, and deficiency in abstract thinking and relevancy of ideas. The authors stated that these characteristics are associated with "haphazardly" introduced words and sentences in conversation. Two of the subjects in the present study demonstrated perseveration and inappropriate random talking.

The three speech characteristics in this study which may be associated with "narrative, compulsive, observational and repetitive speech" reported in the literature occurred in the files of only four of the seven subjects in the present study. This writer interpreted the above mentioned terms as comparable to perseverative speech and inappropriate random talking. Complete definition of terms will be an important component for future studies conducted by speech pathologists.

Ambiguous terminology, anecdotal reporting, and failure to relate observations of fragile X syndrome patients to other mentally retarded patients leave some question remaining about the exact nature of the communicative behaviors of fragile X patients. However, it is the opinion of this writer that there is insufficient evidence that fragile X syndrome patients demonstrate a distinct set of speech and language behaviors which differentiates them from other mentally retarded people. The literature reviewed, the present investigation, and this writer's
clinical experience with mentally retarded children indicate that a comparative study of the speech and language characteristics of fragile X syndrome children will yield little information valuable to clinical management.

The present study may serve as a source of information for speech pathologists interested in fragile X syndrome. People with fragile X syndrome will receive speech and language services in clinical or school settings. Speech pathologists do not play a significant role in diagnosis of the fragile X condition. They may take a more traditional role in lessening the impact of cognitive and communicative disorders on clients and their families (Hecht, et al., 1981).

The speech pathologist may be one of the first professionals involved in a case of delayed development (Matthews, 1957). He or she has a responsibility to encourage clients to focus attention on problems which may accompany speech and language deficiency, and which must be recognized and dealt with (Matthews, 1957). For example, clinicians should be aware of the importance of genetic counseling for families with inherited disorders. Fragile X syndrome is a disorder being diagnosed more and more frequently. Knowledge of the syndrome will facilitate intervention for an increasing number of clients.

Writers of several descriptions of fragile X syndrome children in the literature and in evaluation reports suggested that they were impressed with the communicative behavior of the children. This inves-
tigation indicated that these children exhibit speech and language behaviors like those of other mentally retarded children. The results of this study suggest that comparative investigation of the speech and language behaviors of fragile X syndrome children is not warranted.
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