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ABSTRACT

Analysis of the phonological patterns of two physically normal boys, aged 5 and 8 years, with fragile X syndrome, an X-chromosomal abnormality usually connected with severe to moderate mental retardation, found language features similar to those found in other studies of fragile X speech. Some of these language features are: repetition of initial syllables, perseverations, unintelligibility, and in one or the other child, incoherence, comprehension problems, and fast speech tempo. These linguistic features are similar to speech characteristics of normally-intelligent children with developmental apraxis of speech. Systematic vowel errors and errors in place, manner of articulation, and sonority of consonants were also found. (MSE)

FRAGILE X SPEECH IN FINNISH: PHONOLOGICAL OBSERVATIONS

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1. INTRODUCTION

The term fragile X syndrome is used to refer to a genetic inherited symptom complex which is linked with a marker X chromosome (fra(X), (q27)). In adult male patients this syndrome is usually connected with severe to moderate mental retardation (Vilkman 1984, Simola 1984).

The fragile X condition is not necessarily linked with any physical anomalies. However, some abnormalities such as large ears, high forehead, high arched palate and EEG changes are among the frequently reported clinical features (Tariverdian & Weck 1982). The earliest description of X-linked mental retardation dates back to the 1940's (Martin & Bell 1943). However, the observation of the marker X chromosome was not made possible until the late 1970's when a specific chromosome culture method (e.g. Sutherland 1979) was taken into routine use. In a chromosome examination the marker X chromosome appears as a constriction of the tip of the long arm of the X chromosome (e.g. Silverman et al. 1983). The balancing effect of the other, and normal, X chromosome has been claimed to account for the fact that female patients are only mildly affected or totally unaffected (Turner et al. 1980).

What makes the linguistic examination of the fragile X syndrome very interesting is the assumption that this syndrome may be linked with a specific language disorder (Lehrke 1974, Netley & Rovet 1982, Bender 1983). In intelligence tests of mentally retarded boys, the scores of verbal tasks have been found to be clearly lower than of those which focus on performance. This trend has been claimed to yield evidence in favour of the specific language disorder (Lehrke 1974). Moreover, and to put it more generally, X-linked abnormalities are interesting because they have been suspected to account for the over-representation of boys in the statistics of mental retardation and delayed language development (Arnold 1970).

The definition of the special linguistic features and disabilities of fragile X patients may yield valuable information as regards the development of therapeutical

and diagnostic practices. It has been claimed that even a limited linguistic dysfunction can stop the general development of the child (e.g. McLaughlin & Kriegsman 1980). In this respect, the prevention of this cessation by means of therapy could be regarded as a goal worth aiming at (cf. Vilkman 1984). The development of the therapy of these patients is very important also when one keeps in mind that the marker X chromosome linked mental retardation may be the second most common syndrome caused by chromosome abnormality in boys (Herbst & Miller 1980). It has been estimated that about 10 new cases are born in Finland annually (Simola 1984).

In the long run, the basic aim in the examination of the speech of fragile X patients will be to define what kind of linguistic problems may lay in the background of relative retardation, or cessation, of the development of a child, and, in addition, to examine what possibilities speech therapy can provide for these problems. The linguistic aims of this kind of study, as will be noticed, are mainly psycholinguistic in nature, and the research practices include approaches such as the phonological process analysis (e.g. Magnusson 1983, Nettelbladt 1984) and the word prototype account (e.g. Leonard 1985, Leonard & Brown 1984, Panagos & Prelock 1982). This paper is a preliminary report on some linguistic features of two physically normal fragile X patients, and only tentative information can be presented as regards the above mentioned general aims.

2. PROCEDURE OF THE PHONOLOGICAL ANALYSIS

The linguistic, i.e., phonological analysis of the data was carried out at the University of Joensuu. Several phonological analyses of various language and speech dysfunctions within a linguistic framework have been performed (e.g. Shriberg & Kwiatkowski 1982). Thus, the phonological account adopted here may provide a basis for comparison to other types of language dysfunctions, and also to normal language development, described in a similar manner. In order to define the neture of the expressive language disorder, the results will also be (see chapter 4) compared to some linguistic features of Finnish Broca's aphasics (Niemi 1984, Niemi et al. 1985). In addition, we feel that a phonological analysis is desirable, if not necessary, for the development of therapeutical practices, because phonological dysfunctions can in some cases hamper the overall language development (Leiwo 1977). A more

comprehensive discussion of the results obtained from the analysis of these two cases will be presented in Niemi and Vilkman (to appear).

As regards the content of therapy, a phonological process approach may suggest factors which underlie the child's restricted phonological patterns. The analysis might show what processes result in unintelligibility, and, in addition, disclose which processes give rise homonymy, i.e., show what processes underlie the possible basic phonological word types employed by the child. Thus a phonological analysis can also provide a basis for further analysis, e.g. for a word prototype account (cf. Nettelbladt 1983, Leonard 1985, Leonard & Brown 1985).

In the classification of the sounds as correct or erroneous, structurally-oriented criteria were adopted. A speech sound was scored as correct, if it showed allophonic deviations only. The criterion was thus a phonemic one. In other words, a sound was rated as correct, when it could be judged to be an allophone of its target phoneme, i.e., neither an allophone of some other Finnish phoneme, nor that of a distinctly un-Finnish sound (e.g. w, ə, β, θ).

If we had adopted acceptability criteria defined purely at the level of allophones, nearly all of the analyzed speech sounds ought to have been rated as erroneous. In spite of this "gentle approach" of ours, we found hundreds of phonological errors in the speech of our two patients.

3. SUBJECTS AND MATERIALS

The patients discussed here have been studied at the Phoniatric Department of the Central Hospital of Central Finland. The speech samples were recorded by speech therapists Tuula Pulli, Tuula Jalkanen and Armi Mäkelä. The psychological tests were performed by psychologist Marja-Terttu Kuparinen. To the best of our knowledge, our patients are the youngest fragile X patients ever examined.

3.1. Case 1

Case 1 (C1) was born in 1979. He was for the first time examined at the Phoniatric Department at the age of three because of contact and communication problems. An examination was performed to find the marker X chromosome, because his younger brother also showed developmental retardation. The patient learned to walk at 1:2, and the first words were produced approximately at the same

age. From the age of four onwards the patient has attended speech therapy, which has not been without problems because of his hyperkineticity and his inability to concentrate.

C1 was 5:4 at the time of the recordings. His speech sample shows that he can separately produce all Finnish speech sounds. However, his speech exhibits perseveration, clutter, and repetition of initial syllables. Because of these features, spontaneous speech is often unclear and also exhibits phonological and morphological errors. In addition, his speech is often incoherent and wildly associative. Speech-motor evaluation exhibited clumsiness especially with lips. There was also some drivelling. Psychological testing has been particularly problematic because C1 has difficulties in persisting in tasks. Moreover, the patient has clear comprehension problems in verbal tasks.

Performance IQ (Leiter) of C1 was 69 (total IQ and verbal IQ could not be obtained). The patient's intelligence was thus at borderline level. (Note that at the age of three C1 rated 83 in another test which emphasizes performance (M-P).) Physically C1 is in good proportion and his rough and fine motor activities are within normal limits. The patient's hearing is normal, also. The marker X chromosome is found in 20 % of examined cells. C1 goes to kirdergarten and gets speech therapy rehabilitation. Moreover, periods of therapy at the Phoniatric Department are part of his therapy program.

3.2. Case 2

The patient was born in 1976, and for the first time he was taken to the Phoniatric Clinic at the age of 2:2 to be evaluated due to developmental language retardation. The marker X chromosome was found in conjunction with thorough evaluations of retardation. C2 learnt to walk at 1:7 and the development of his rough motor abilities has been within the limits of normal, whereas some clumsiness has been apparent in his fine motor actions. The patient's oral motor skills are poor and drivelling has appeared continuously. Speech therapy was started when C2 was three years old. Hyperactivity, concentration difficulties, low anxiety threshold in tasks and shyness has made the therapy very difficult. In intelligence tests (WPPSI) C2 received a total IQ of 63. Performance IQ was 62 and verbal IQ 56. C2 was thus rated as mildly mentally retarded. The age of the patient was 7:6 at the time of the clinical evaluation. Of these two patients, C2 is clearly the more retarded one.

C2 was 8:0 at the time of the recording of the speech sample. As regards his present speech, typical audible features have included repetition of initial syllables,

fast speech tempo, and clutter, all resulting in difficulties in intelligibility. In addition, C2's speech includes a large amount of perseverations. The patient can produce all Finnish sounds separately and he often uses grammatically correct complex utterances. He produced his first words about the age of two, but two-word utterances were not employed until the age of four.

The patient is physically normal, his ears are within the limits of normal; his face is longish and narrowish. The marker X chromosome was found in 30 % of the cells examined. At the moment, the patient attends a special school for aphasic children.

4. RESULTS AND DISCUSSION

4.1. General

A recently published preliminary report (Paul et al. 1984) on the linguistic proficiency of three 10-13 year-old fragile X patients states that these individuals show a specific verbal dyspractic disability. In addition to mild to moderate mental retardation, the reported similarities between fragile X patients include the following linguistic features: 1) poorer performance on production than on receptive language or nonverbal tasks, 2) poor intelligibility in connected speech despite good performance on isolated words in articulation tests, 3) the use of a very limited number of phonological simplification processes, 4) poor performance in repetition tasks, 5) disfluency, e.g. prolongations and repetitions of sounds and syllables.

As regards our patients, they exhibit language features very similar to those reported in other studies of fragile X speech: they both showed repetition of initial syllables, perseverations and unintelligibility. In addition, C1 exhibited such features as incoherence and comprehension problems in verbal tasks and C2 showed fast speech tempo. These linguistic features are similar to the speech characteristics of children of normal intelligence with developmental apraxia of speech.

4.2. Vowels

To generalize, the pathological phonology of our patients seems to be rather systematic, i.e., failures occur in a systematic manner. As regards vowel errors, the following trends can be mentioned:

1) Back V ==> front V; i.e., there is a clear tendency of fronting; this trend seems to be the strongest one among the vowel errors.

2) Close V ==> ∅; i.e., close vowels tend to be easily omitted. The total amount of vowel errors was 70, 37 of which were omissions.

3) V: ==> V; disappearance of quantity opposition. A rather rare error-type; this is found also in Broca's aphasics. The rareness of this feature can be taken to imply that speakers of a quantity language like Finnish are constrained by the short-long boundary even in the face of severe language impairment (cf. Niemi et al. 1985).

4) Labiality is resistant to error. Also Broca's aphasics seem to exhibit this feature (cf. Niemi et al. op.cit.). This trend is somewhat surprising, because the oral motor skills of both of the patients are rather poor.

4.3. Consonants

In this connection, the most important trends concerning place, manner, and sonority of consonants will be briefly discussed.

As regards place of articulation, dentals seem to be the most sensitive groups for substitutions. The direction of errors of C1 is mainly towards /h/and velars, and in C2 towards /h/, velars, and labials. The basic difference between the two cases seems to be in the amount of labiality errors. Note that the high rate of /h/ can most likely be explained as an error of manner, and not as one of place.

Omissions follow a similar pattern in both cases: dentals are very easily omitted, and velars are the next most frequently omitted group as far as place of articulation is concerned.

As for manner of articulation, the rank order of sensitivity for change is the following: /r/ > SIB > STO > NAS > SPI > /l/ > /j/. /r/, sibilants and stops attract most of the errors, whereas /j/ is always j-like. Laterals, spirants and nasals include relatively few errors.

As regards omissions in relation to manner of articulation, nasals are easily omitted by C1, and stops are not very far behind. C2 is characterized by the omission of stops while other classes show a few errors only.

It seems that substitutions are the most common phonological dysfunction type both in fragile X syndrome and in Broca's aphasia. The proportion of omissions and additions is rather even in aphasia, whereas fragile X patients exhibit only a few additions. (Cf. Niemi et al. 1985.)

Changes in the sonority of consonants were observed to take place along the following continuum:

<------ <-----

STO - FRI - LIQ - NAS - (/j/)

<----------

The arrows above indicate the general trends of change in sonority. It can be seen that stops are usually substituted for fricatives, and fricatives are substituted for liquids and nasals. No sonority changes concerning /j/ could be observed.

In addition, a "loss and gain" analysis was performed as far as both place and manner of articulation are concerned. The results of this analysis are shown in Figures 1 and 2. In these figures the values, i.e., bars above the 0 %-line indicate how often a substitution, a phonological process, took place towards a certain category of place or manner of articulation. For example, the move could be from manner X towards spirants (X ==> SPI). The bars below the zero-line indicate how often the move was away form a certain category, e.g., SPI ==> X.

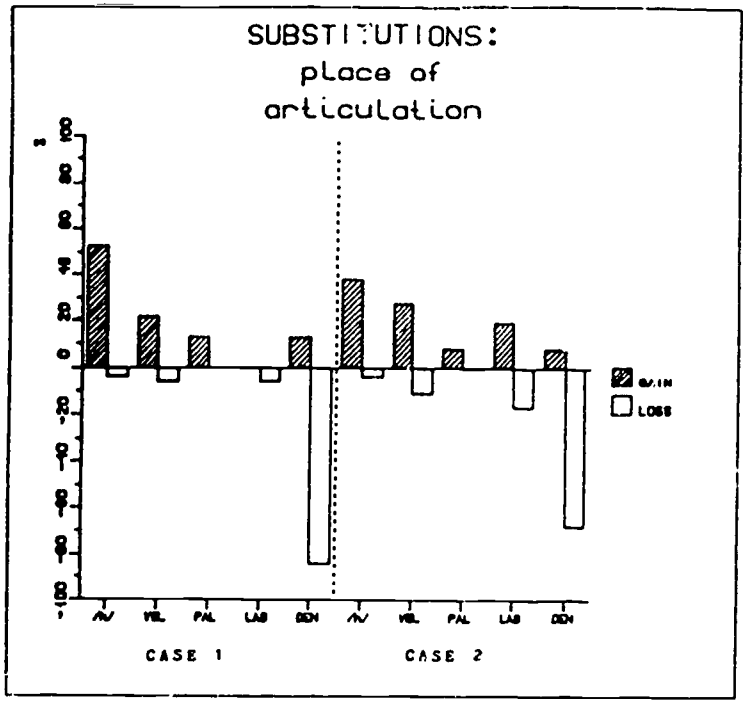

Figure 1.

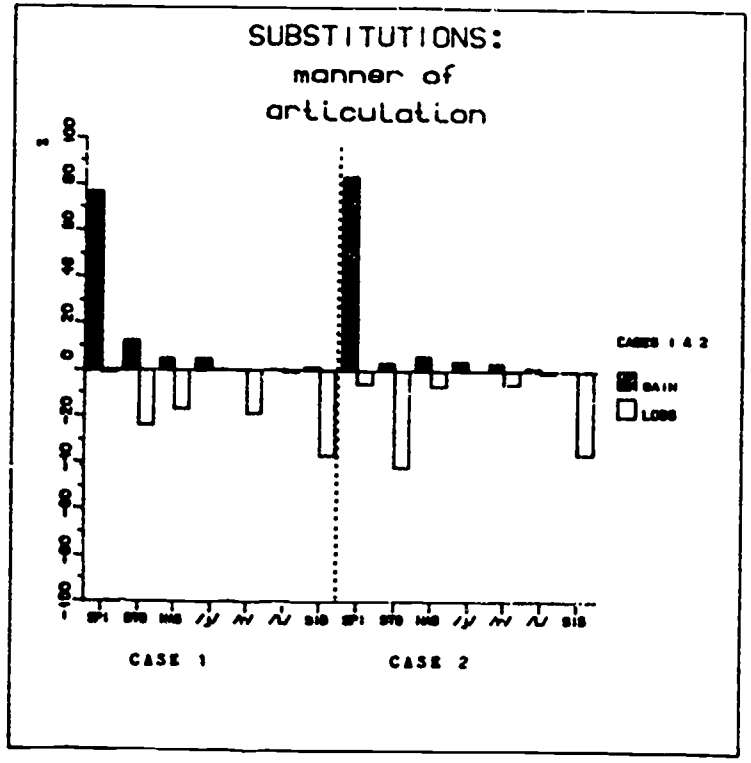

Figure 2.

As for place of articulation, the general trend is clearly rather identical in both cases. The older patient (C2) exhibits perhaps a more stable distribution of substitutions. One could assume that the distribution becomes more and more stable in the course of the development and growth of the patient. The direction of substitutions is mainly towards /h/ in both cases; this is most likely an error of manner of articulation, however. Dentals, on the other hand, seem to be the most unstable category of place with these patients.

Spirantization (h-zation) seems to be the major phonological process concerning manner of articulation. In addition, one can note that sibilants and stops are the most prone categories for substitution with these fragile X boys.

As fas as future research, the word prototype analysis may yield useful linguistic insights and, in addition, provide a basis for the development of diagnosis and therapy of fragile X patients. One additional future direction for research might be the comparison of the results obtained here to the linguistic features of Down's syndrome patients.

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