



# Fragile X

-

( , , , )  
(가 )  
( )  
( )

. . . . . **Fragile X** . . . . .  
., 1998, 3 , 160- 174. Fragile X Down . F  
- 가 . 6-30 6 - 가  
. 가  
, 2-4 , 1-3  
, 3  
3-37 % 가 ,  
가가 4  
. 6 5 가  
, 1 - 가 Fragile X가 가  
Fragile X - 가

## I.

Fragile X 1943 Martin Bell X -  
Down

$1/1000 - 1/1250$ ,  $1/2000$  (heterozygote)  $1/700$   
(Webb, 1991; Laxova, 1994).

( ) 가 . 122

2 Fragile X site

1.6 %

7.7 %

Fragile X

X- 가 Xq27.3 FMR- 1 가  
 (Turk, 1992; Laxova, 1994). 50 %  
 99 %  
 50 %  
 (premutation) (Normal  
 Transmitting Male)  
 가

1. Fragile X

, , .  
 , , , 가 , , , ,  
 가 가 ( . . ,  
 1997).  
 . 10- 15 가  
 , (Hagerman  
 et al., 1989; Turk, 1992).

2. Fragile X

Fragile X  
 (Turk, 1992).  
 (auditory short term memory), (auditory sequencing) 가  
 (Paul et al., 1984), /  
 (Bergman et al., 1987; Hanson, Jackson & Hagerman, 1986).  
 (Spinelli et al., 1995), 가  
 가 .

1992). (Grigsby, Kemper & Hagerman, Down  
 (Miller, 1987). Carpenter, Leichtman and Say (1982) Fragile X  
 (Sudhalter, Maranion & Brooks, 1992)  
 가 가  
 Down  
 (Ferrier et al., 1991; Jorgensen et al., 1984; ,  
 1992) 14- 16%가 Fragile X  
 Lehrke (1972) 가 가  
 가 X - 가  
 (posterior vermis) 가 (Reiss et al., 1991)  
 가

### 3. Fragile X

Fragile X  
 (prosody) (Paul et al., 1984; Newell,  
 Sanborn, & Hagerman, 1983). (litany speech)  
 (Turner, Daniel & Frost,  
 1980). . Fragile X  
 (dyspraxia), (dysfluency),  
 (incomplete sentences), (revision behavior), (palilalia),  
 (cluttering) (Hanson et al., 1986; Paul et al., 1984; Spinelli et al., 1995),  
 , (perseveration) (Turk, 1992).  
 Fragile X  
 Fragile X  
 (Ferrier et al., 1991)  
 (Paul et al., 1987).

Ferrier

et al. (1991)

Fragile X

가

Fragile X

가

Fragile X

6

가가

## II.

### 1.

122

Fragile X

2

가

1

2

가

Fragile X

Fragile X

1

가

. 6

6 - 30

13.50

9.12

6

5

3

가

0.06-48 (VMI

KEDI-WISC )

, 2

41, 48

2

, 3

### 2.

2

가

가

, 5

가

SONY video camera recorder CCD-TR75

1

Marantz PMD430

가

(

, 1995)

(Preschool Language Scale, 1979;

1994)

( , 1994)

( ( . , 1997; < - 1> ) . 가 / / , , , .

### 3.

. , , .  
(vocalization)  
(unintelligible utterance), 가 (elicited utterance), / / .

. 가  
(sound transposition), (searching behavior)  
가 가 (Spinelli et al., 1995).  
(dysfluency) , , , ,  
가 (palilalia) , 가  
가 (Newell et al., 1983)  
(cluttering)  
가  
(incomplete articulation) ,  
가

## III.

### 1.

6

S<sub>1</sub> - S<sub>6</sub>

가. /

< - 1 >

S <sub>1</sub> (30;10)	2	1 7 ; 1 6
S <sub>2</sub> (18;5)	3 -3 5	3 1 ; 2 1
S <sub>3</sub> (11;11)	4 6 -11	3 4 ; 3
S <sub>4</sub> (8;8)		
S <sub>5</sub> (8;2)	2 -2 5	2 ; 2 4
S <sub>6</sub> (6;7)	4 -4 5	3 ; 2 10

(descriptive)

S<sub>1</sub> 158 , S<sub>2</sub> 108 , S<sub>3</sub> 127 , S<sub>4</sub> 19 , S<sub>5</sub> 104 , S<sub>6</sub> 252 .

S<sub>3</sub>, S<sub>5</sub>, S<sub>6</sub>

< - 2 >

(entity)

가 가

가

3가

(%)

(< - 3 >

).

(1997)

(acknowledgment) (Aram & Nation, 1982)

가

< - 2>

S <sub>1</sub> (30;10)	
S <sub>2</sub> (18;5)	
S <sub>3</sub> (11;11)	/ -
S <sub>4</sub> (8;8)	
S <sub>5</sub> (8;2)	/ - - -
S <sub>6</sub> (6;7)	/ - - - - - - -

< - 3> 가 (%)

S <sub>1</sub> (30;10)	39.5	42.0				10.8
S <sub>2</sub> (18;5)	50.5	13.1			10.3	
S <sub>3</sub> (11;11)	15.1	68.3			4.0	
S <sub>4</sub> (8;8)		63.2	10.5			21.1
S <sub>5</sub> (8;2)	42.7	32.0		5.8		
S <sub>6</sub> (6;7)	26.1	18.9	13.7			

( , , , ) ( , , ) 가 가  
 ( , , , , ), ( , , , 가  
 ), ( , , , ) 가 . ( ,  
 , , , ) . 1-2  
 가 , < - 4> .

, S<sub>1</sub> 46.2 %, S<sub>2</sub> 25.0 %, S<sub>3</sub> 13.2 % 가

가 S<sub>4</sub> 0%, S<sub>5</sub> 39.4%, S<sub>6</sub> 13.9% .

가 가 < - 5> .

< - 4>

S <sub>1</sub> (30;10)	/
S <sub>2</sub> (18;5)	/
S <sub>3</sub> (11;11)	/
S <sub>4</sub> (8;8)	
S <sub>5</sub> (8;2)	/
S <sub>6</sub> (6;7)	/

< - 5> 가 (%)

S <sub>1</sub> (30;10)	59.5			7.6	18.4	
S <sub>2</sub> (18;5)	56.0			16.5	22.0	
S <sub>3</sub> (11;11)	69.4	9.9				5.8
S <sub>4</sub> (8;8)	15.8	47.4	21.1			
S <sub>5</sub> (8;2)	52.9	24.0	11.5			
S <sub>6</sub> (6;7)	61.9		6.2	8.3		

2.

가

< - 6> (%) .



(vocal intensity) 60 cm

가 4 가가

< - 6> (%)

S <sub>1</sub> (30;10)	1.2	0	5.0	57.6
S <sub>2</sub> (18;5)	4.6	0	3.7	89.0
S <sub>3</sub> (11;11)	2.5	0	11.6	95.9
S <sub>4</sub> (8;8)	42.1	0	36.8	73.7
S <sub>5</sub> (8;2)	29.8	0	27.9	81.7
S <sub>6</sub> (6;7)	9.9	0	6.0	46.0

가 가

Fragile X

( : 가 )

(shyness)

가

5

(impulsivity)

4

(immediacy)

#### IV.

Fragile X

. Fragile X

( : )

Fragile X

-

가

Fragile X

2-4

가 6

30

가

가

(Miller, 1987; Paul et al., 1987)

가

Lehrke (1974), Hagerman and McBogg (1983) X

(Ferrier et al., 1991; Hanson et al., 1986; Spinelli

et al., 1995; Sudhalter et al., 1992)

가

1-2

19-252

2-3

3

2-3

1-2

가

가

19

S4

5

13.9-46.2 %

Fragile X

1.2 % 42.1 %

3.7 % 37.8 %

40 %

95.9 %가

가,  
 4 가가 가 Fragile X 가  
 가  
 , 6 1 Fragile X  
 - 가 가 가가  
 가 가  
 , 20 %가  
 50 %가 가 가 가가  
 - 가 가  
 가 Fragile X 가 Fragile X  
 가 , ,  
 가 .

(1994). 『 - 』.  
 (1995). : .  
 (1992). X . *Korean J Child & Adol Psychiatr*, 3,  
 147- 157.  
 (1997).  
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< - 1 >

			(%)	
	가			

## **ABSTRACT**

### **Speech and Language of People with Fragile X Syndrome**

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(Samsung Medical Center, Karam Neuropsychiatric Clinic,  
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Fragile X syndrome is the second most common cause of mental retardation next to Down syndrome. This study was attempted to describe language and speech characteristics of patients with fragile X. The subject group of this study includes six patients with full mutation fragile X. Full speech-language evaluations were conducted on each subject. In addition, language samples were analyzed in the aspects of semantics and pragmatics. Speech was observed with respect to dyspraxia, dysfluency, palilalia, and incomplete articulation. The results of language analysis revealed that all of the subjects were collected and severely disordered: (1) vocabulary level was compatible with that of two to four-year-old children, (2) level of receptive and expressive language was that of one to three-year-old children, (3) their utterances, consisting mostly of one word, were limited to the semantic categories of "entity" or "deictic", and (4) pragmatics was also extremely defective in that a large part of their utterances was responses to yes/no questions with sparse identifications. The results of the speech evaluation showed that (1) incomplete articulation was the most dominant feature, (2) palilalia was noticed quite frequently, (3) symptoms of dyspraxia varied rather widely spanning 3-37 % of their utterances, and (4) dysfluency was not found at all. On the basis of the results, we concluded that a patient with fragile X syndrome could be diagnosed exclusively with a speech-language evaluation instead of expensive neuro-behavioral tests such as a cytogenetic test.

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