

## 9번 염색체 전위를 지닌 환아들의 정신과적 특성\*

- 예비적 연구 -

### PSYCHIATRIC CHARACTERISTICS OF CHILD PATIENTS WITH INVERSION OF CHROMOSOME 9 - A PRELIMINARY STUDY -

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연구 목적 : 9번 염색체 전위를 지닌 환아들의 정신과적 특성을 알아보고, 9번 염색체 전위와 정신과적 특성 간의 관련성을 규명하는 데 목적이 있다.

방 법 : 1) 1984년 1월 1일부터 2000년 12월 31일까지 서울대학교병원 소아정신과에 내원한 9번 염색체 전위를 지닌 환아 45명 (Korean Child Behavior Check List, K-CBCL)을 대상으로 하였다.

결 과 : 1) 9번 염색체 전위를 지닌 환아 45명 중 (47.3%), (23.6%), (17.0%), (35.8%), (31.3%), Fragile X (23.9%), (20.9%)을 동반하였다.

2) K-CBCL 점수는 정상 범주에 속하는 환아는 17명 (37.8%)이었다.

결 론 : 9번 염색체 전위를 지닌 환아는 정상 범주에 속하는 환아보다 정신과적 특성이 더 심한 경향을 보였다.

중심 단어 : 9번 염색체 전위, 정신과적 특성

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## 서 론

9 IQ 63.0, SQ 69.0  
 9 (pericentric inversion) 17.6  
 (human karyotype) 가 9  
<sup>1)2)</sup> 9 가  
 (normal variant) 가 . Gard-  
 ener Sutherland<sup>3)</sup> 9  
 9 가 (centromere)  
 (long arm) (short arm)  
 (breaking point) 180  
 9  
 4-6), 7), 8),  
 9)  
 2)10)12)  
 9 가 . Serra <sup>8)</sup>  
 0.85% Hsu <sup>11)</sup>  
 0.26% 가  
 . Yamada<sup>12)</sup>, Ko <sup>13)</sup>, Teo<sup>14)</sup>  
 1.65%, 1.20%, 1.20%  
 8) 가  
 12) 가  
 15)  
 9 1.70%  
 25.0% 가  
 (23.1%) (7.7%)  
<sup>16)</sup>  
 가  
 9  
 가 (2.50%)

## 연구 방법

### 1. 연구 대상

1984 1 2000 12  
 가 9  
 가

**Table 1.** Socio-demographic characteristics of study subjects

Variables/ Group(No)	Inv(9) (N=12)	Control(N=45)
Sex(No.)	M(8)/F(4)	M(26)/F(19)
Mean age(S.D.)*	10.42(5.33)	9.98(0.78)
SES(%)**	High(0) Middle(11) Low(1)	High(1) Middle(44) Low(0)
Father's education(No.)	University(8) High school(2) Middle school(1) Elementary school(1)	University(30) High school(15) Middle school(0) Elementary school(0)
Mother's education(No.)	University(6) High school(3) Middle school(1) Elementary school(2)	University(23) High school(20) Middle school(2) Elementary school(0)

\* : S.D. : Standard Deviation

\*\* : SES : Socio-economic-status

<sup>2</sup>-test for Sex, SES, Father and Mother's degree AND Student's t-test for age





**Table 4.** Developmental history

	Inv(9)	Control	p value
Social smile, head control, crawling, sitting, standing, walking(%)	Early(45.5%) At the right time(27.3%) Late(27.3%)	Early(26.8%) At the right time(68.3%) Late(4.9%)	NS
Babbling, saying 'Mom' & 'Dad' (%)	Early(27.3%) At the right time(18.2%) Late(54.5%)	Early(17.1%) At the right time(78.0%) Late(4.9%)	0.001*
Used real sentences with grammatical function for the first time	52.11 ± 37.82 <sup>a</sup>	30.97 ± 9.18	NS
Completed toilet training	41.55 ± 28.98	25.38 ± 12.48	0.021**

\* : statistically significant in Fisher's exact test ('Early' and 'At the right time' considered as one group)

\*\* : statistically significant in Mann-Whitney U-test      <sup>a</sup> : values are mean ± S.D.      NS : Not significant

가 2~3 가 22)23) 가 . Samonte<sup>24)</sup> (fluorescence in situ hybridization, FISH) (breakpoint) 4가 9 Ramesh Verma<sup>25)</sup> 가 ) (52.11 ) (30.97 3가 9 p<0.05 가 9 0.05 0.1 가 4-6) 가 0.05 (p<0.1) 10)12)26-28)가 가 (p<0.01) 가 가 9 가 41.55 (25.38 ) 가 9 가 (p<0.05).



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**Objectives :** Few studies have examined the psychiatric properties or child developmental problems associated with inversion of chromosome 9. The purpose of this study is to examine the psychiatric properties of child patients who have inversion of chromosome 9, focused on behavioral problems and child developmental problems like motor or language developmental delay, intellectual impairment, and growth retardation.

**Methods :**

1) The authors examined the cases referred for cytogenetic examination from 1984 to 2000 at Seoul National University Hospital in Korea. The cases with the examination result of inversion of chromosome 9 were collected and informations about the departments which referred and the main reasons for referral were also checked.

2) 12 child subjects with inversion of chromosome 9 and their parents underwent psychiatric interview and parent questionnaire (child and adolescent past history questionnaire, CBCL). 45 normal students whose sex and age were matched to patients were selected as a control group.

**Results :**

1) There were 165 cases of inversion of chromosome 9. The major departments which referred were Obstetrics and Gynecology (47.3%), Pediatrics (23.6%) and Child and Adolescent Psychiatry (17.0%). The major reasons for referral from the Pediatrics and the Child and Adolescent Psychiatry department (67 cases total) were intellectual impairment (35.8%), language or motor developmental delay (31.3%), suspected Fragile X syndrome (23.9%), and growth retardation (20.9%).

2) Compared to normal control group, the rate to be included in the clinical range with regard to the social problems profile was higher in patient group according to the CBCL results. The patient group had language and motor developmental delay.

**Conclusion :** There is a possibility of inversion of chromosome 9 to be associated with child developmental problems or behavioral problems. This study is the first approach to evaluate the developmental aspects associated with inversion of chromosome 9.

**KEY WORDS :** Inversion of chromosome 9 · Development · Behavioral problems.