

정신지체와 자폐장애를 보이는 Cornelia De Lange 증후군 1예

A CASE OF CORENELIA DE LANGE SYNDROME WITH MENTAL RETARDATION AND AUTISTIC DISORDER

송정은*† · 김세주* · 최낙경*

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요 약 : Cornelia de Lange

Cornelia de Lange
가
Cornelia de Lange

6 4

가

가

4

Cornelia de Lange

중심 단어 : Cornelia de lange

4), 5)

서 론

Cornelia

de Lange

Cornelia de Lange

1933 de Lange¹⁾

10000 1

Brachmann

⁶⁾ Cornelia de Lange

가

3q

가

⁷⁾

Brachmann - de Lange

²⁾

가

1967

³⁾

1

99%

⁸⁾

*
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Cornelia de Lange

3
2 9

가 , 가 1

5)

2. 발달력 및 가족력

2 1

9)(10)

25 , 29

가

39

Cornelia de

Lange

2.5kg(3 percentile)

46cm(3 percentile)

증 례

1. 주소 및 현 병력

6 4

가 가

8 , 14 , 20 , 24

가

가 3 ,

“ , 가 ”

15

24

7

. TV , 4~

45

3 , 6

5

. 20

1

24

1

가

가 , 6 5

. 5

22

가

1

12

3

10

14

가 1

. 36

21 ,

13

1

27

12

가

1 6

, 27

3. 정신상태검사 및 신체이학적 검사

104cm(<3 percentile),
20kg(50 percentile), 53cm(>97 percentile)

가

(Fig. 1).



Fig. 1. The characteristic face of Cornelia de Lange syndrome.



Fig. 2. Hypertrichosis in back.

(Fig. 2).

가

가

가

“ ~, ~ ”

1 6 , 24

가

32.5

DSM -

가

고 찰

Cornelia de Lange

de Lange IQ 53(30~85)

30~60%

가

가 ¹¹⁾ 50%

. 4

Cornelia de Lange

53%가

33%가

¹²⁾

가

가

Cornelia de Lange

가

가

Cornelia de Lange

References

13)14)
 Cornelia de Lange
 가 가
 15)
 가
 16) 49 Cornelia
 de Lange
 , , ,
 가
 가
 53%가 ICD - 10
 가
 , , ,
 ,
 DSM - IV
 Cornelia de Lange
 가
 12% 23%
 25%
 2)
 , , ,
 ,
 Cornelia de Lange
 ,
 Cor-

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Cornelia de Lange syndrome is a dysmorphogenic disorder characterized by multiple congenital abnormalities, mental retardation, growth retardation and neurodevelopmental abnormalities. Diagnosis for the Cornelia de Lange syndrome is dependent on the clinical observation because neither definite biological marker nor definite chromosomal abnormality have been investigated. Clinical observation is important for the diagnosis, so we report a case of Cornelia de Lange syndrome with mental retardation and autistic disorder.

The patient is a 6-year old girl. Her motor development and language development have been delayed. She could say no meaningful word and understood simple command partially. She showed poor eye contact and poor emotional interaction. Social interaction was impaired and she showed stereotypic behaviors. Thus we diagnosed her as mental retardation with autistic disorder.

She had vesicoureteral reflux, frequent upper respiratory infection and pneumonia. She had experienced febrile convulsions 4 times. She had short stature, confluent eyebrows, long eyelashes, and upturned nose with anteverted nostrils. She also showed low hairline and hypertrichosis in body and extremities. Her finger was short.

In this case, we diagnosed Cornelia de Lange syndrome by her characteristic face, hypertrichosis and medical and behavioral problems that were frequently showed in this syndrome.

KEY WORDS : Cornelia de lange syndrome · Mental retardation · Autistic disorder.