소아재활

게시일시 및 장소 : 10 월 19 일(토) 08:30-12:30 Room G(3F)

질의응답 일시 및 장소 : 10 월 19 일(토) 11:00-11:30 Room G(3F)

P 3-100

Pediatric tetrasomy 18p presenting as a spastic cerebral palsy: a case report

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Introduction

Tetrasomy 18p syndrome is a genetic syndrome caused by an isochromosome consisting of chromosome 18 short arms. 18p isochromosome causes various symptoms including developmental delay, intellectual disability and abnormal muscle tone. This tetrasomy 18p case is about a patient with developmental delay and increased spasticity, previously diagnosed as a spastic diplegic cerebral palsy in another hospital.

Case report

An eight-month-old female presented with a delayed milestone. Because she could not able to creep and had a severe spasticity of both legs, clinically cerebral palsy (CP), specifically spastic diplegic type was suspected. She was born vaginally at 41 weeks of gestation with weight of 2.6kg (5-10%) and was the second child of healthy parents. At the time of birth, her Korean father was 24 years old, her Malian mother was 26 years old. At 8 months of age, the body weight was 7.2 kg (10-25%), and the height was 72.6 cm (90-97%), and the head was 41 cm compatible with microcephaly. Bayley Scales of Infant Development II showed delayed motor development compatible with an age of 4 months. She was classified in Gross Motor Function Classification System level V. On physical examination, microtia and smooth philtrum were also observed (Figure 1). Spasticity, as well as hypertonus and opisthotonus, involving both legs symmetrically was observed in bilateral hip adductors (grade 2), hamstrings (grade 1+), and heelcords (grade 2) by modified Ashworth Scale. On brain magnetic resonance image (MRI), volume loss of periventricular white matter and corpus callosum in parietal area was shown (Figure 2), although this was not a typical finding of periventricular leukomalacia usually shown in CP. High-resolution chromosome study and multiplex ligation-dependent probe amplification (MLPA) tests were conducted. In chromosome study, small metacentric marker chromosome was observed in all of blood metaphase cells (Figure 3). The MLPA results showed peak ratio at 18p11.32 region, which suggested tetrasomy 18p (+i(18p)). Fluorescence in situ hybridization (FISH) analysis was conducted using CEP 18 (D18Z1) SpectrumOrange probe (18p11.1-q11.1) to confirm the diagnosis. Alike all the other 18p

isochromosome cases identified to date which were monocentric, and most of them being de novo cases, the chromosome study of parents were revealed to be normal.

Conclusion

CP is a neurological disorder, including developmental delay caused by a nonprogressive lesion of developing brain. Recently, as genetic test becomes more accessible, the comorbid genetic causes of delayed development more have been identified. Especially, the identification of pathogenic genes or chromosome abnormality is imperative in surveillance of complications in related syndromes and prediction of clinical course. Therefore, genetic testing should be considered in patients with subtle minor physical anomalies or with equivocal MRI findings.

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Figure 1. Dysmorphic features of the patient, including microtia and smooth philtrum

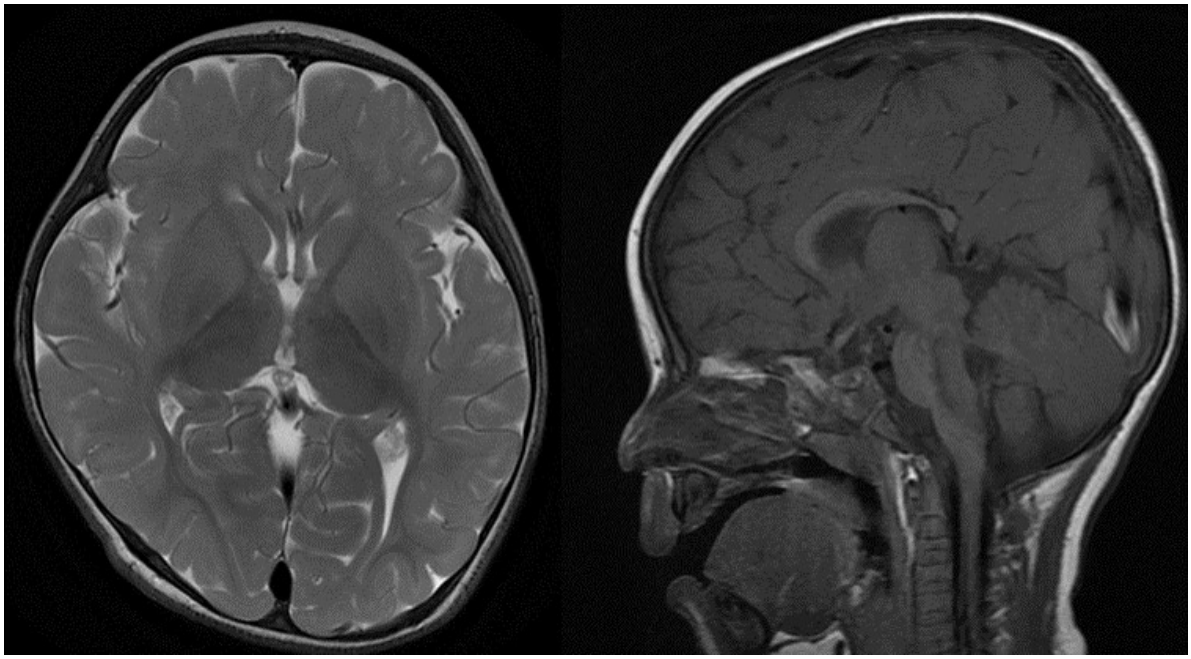

Figure 2. Brain MRI (T1-weighted, axial (left) and sagittal (right). Suspected decreased white matter volumes in parietal areas, with small splenium and posterior body of corpus callosum

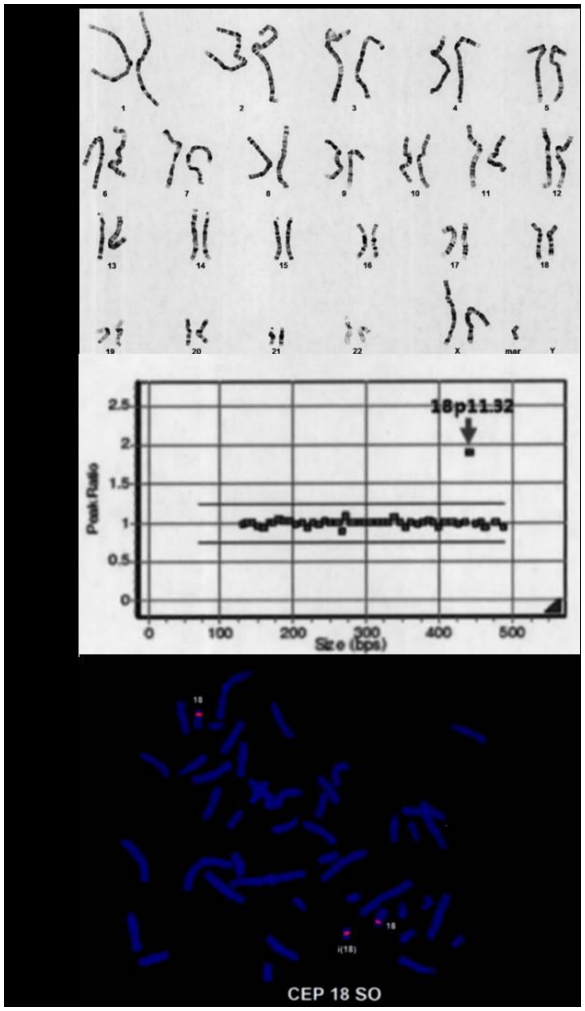

Figure 3. Genetic testing results. (a) High-resolution chromosome study (b) MLPA results (SALSA P070 subtelomeric probemix) (c) FISH using CEP 18 (D18Z1) SpectrumOrange probe. Results summary: 47,XX,+mar.ish i(18)(p10)(D18Z1+)