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)

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Developmental delay in a child with 20q11.2 microdeletion syndrome : A case report

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Introduction

Interstitial microdeletion of the chromosome 20q region, namely the 20q11.2 microdeletion syndrome, is a rare disease. Clinical findings include intellectual disability, motor developmental delay, craniofacial dysmorphism, anomalies of the extremities and feeding difficulties. Associated prenatal growth retardation, gastrointestinal, heart and eye anomalies have also been reported. In this article, we report a case of 20q11.2 microdeletion syndrome patient with developmental delay.

Case Report

A 20 months old girl visited our rehabilitation department because of developmental delay. She was born at 40th week of gestation by normal spontaneous vaginal delivery as the third child of unrelated nonconsanguineous parents. Her birth weight was 2900g (10-25th percentile(%ile)). The child could walk while grabbing onto things, but was unable to ambulate on her own. Otherwise, she did not have abnormal physical examination findings. While undergoing rehabilitation at another hospital, she did not return for follow up. At the age of 45 months, she re-visited our hospital for further developmental evaluation. At the time, her height was 88.1cm(<3rd %ile), weight 11.2kg(<3rd %ile), and occipital-frontal circumference 47.3cm(3-10th %ile). On Bayley Scales of Infant and Toddler Development – 3rd Edition; cognition 85(16th %ile), language 74(4th %ile), motor 91(27th %ile), social-emotion 100(50th %ile), general adaptive composite 65(1st %ile). On Preschool Receptive & Expressive Scale(PRES); reception at the level of 31 months(2nd %ile), expression at 23 months(<1th %ile). Again, she was lost for follow up since then. When the patient became 6 years old, she returned to our department because of scoliosis. On physical examination, her height and weight were 106cm(<3rd %ile) and 21kg(25-50th %ile). She also revealed a crouching gait pattern when asked to walk. Further investigations showed thoracic spine scoliosis(Cobb's angle 14.1) on whole spine x-ray and both hip joint dislocations on lower extremity scannogram. The MRI results of the child's brain showed diffusely enlarged ventricles and atrophic changes. The patient's array comparative genomic

hybridization(CGH) test showed approximately 1.9Mb deletion within chromosome 20(arr 20q11.22-q11.23). Consequently, she was diagnosed to have 20q11.2 microdeletion syndrome. She underwent surgery for hip dislocations last year, and is currently in a walker assisted gait while undergoing rehabilitation for ambulation.

Conclusion

To the best of our knowledge, this is the first case of a 20q11.2 microdeletion syndrome patient to be reported in the Korean population. We also showed that chromosomal microarray can be a useful tool for identifying causative pathogenic variants in patients with developmental delay.