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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Chiari malformation with syringomyelia associated with otopalatodigital syndrome : A case report

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Introduction

The term otopalatodigital syndrome spectrum disorders is an umbrella category that includes four phenotypically related conditions, otopalatodigital syndrome types 1 and 2(OPD1 and OPD2), frontometaphyseal dysplasia(FMD), Melnick–Needles syndrome(MNS), and Terminal osseous dysplasia with pigmentary skin defects(TODPD). FLNA is the only gene in which pathogenic variants are known to cause the otopalatodigital spectrum disorders. We report the first case of a patient with Chiari malformation and syringomyelia in otopalatodigital syndrome spectrum disorders in this study.

Case report

A 12-year-old boy visited our clinic complaining of back pain in March 2019. He had suffered from back pain without in radiating pain of lower extremity for two years. He had bilateral sensorineural hearing loss and diagnosis of intellectual disability due to unknown origin. In addition, he underwent surgery twice because of the contracture of left 5th finger. In addition, he underwent surgery twice because of the contracture of left 5th finger. As a family history, he had two cousins who were not diagnosed any disease with similar appearance and morphology. On physical examination, he showed contracture of multiple proximal interphalangeal joints in both hands. In addition, his face represented full cheek and micrognathia. Furthermore, scoliosis and hypodontia were also noted. On radiologic findings, his skull x-ray exhibited diffuse skull thickening and absence of frontal sinus. Also, there was scoliosis in whole spine x-ray and mild bowing figure in long bone x-ray in upper and lower limbs. In brain magnetic resonance imaging, there was cerebellar tonsilar inferior herniation. In whole spine magnetic resonance imaging, huge hydrosyringomyelia from C1 to T12 level was confirmed with Chiari I malformation. In exome next-generation sequencing (NGS) test, heterozygous missense mutation in FLNA gene was confirmed. As a result, this patient was diagnosed with otopalatodigital syndrome spectrum disorders.