■ Case Report

A Case of a Girl with Arnold–Chiari Type 1 Malformation with Precocious Puberty

Min Sun Kim, Pyoung Han Hwang, Dae–Yeol Lee*

Department of Pediatrics, Chonbuk National University Medical School, Research Institute of Clinical Medicine of Chonbuk National University–Biomedical Institute of Chonbuk National University Hospital, Jeonju, Korea

A small percentage of individuals have the neurological anomaly of central precocious puberty (CPP). Common neurologic causes of CPP include a tumor or congenital lesions. Although Arnold–Chiari malformation can be caused by congenital or acquired causes, it is unusual in patients with CPP. We present the case of a girl aged 4.5 years who complained of breast budding. Her neurological examination and growth pattern were normal. She had no endocrinological abnormality, except for true precocious puberty. We performed brain magnetic resonance imaging, which showed an Arnold–Chiari type 1 malformation. Currently, this case represents the youngest girl who exhibited both Arnold–Chiari type 1 malformation and precocious puberty. Furthermore, it is likely that there is a meaningful association between the brain lesion and precocious puberty in this case.

Keywords: Precocious Puberty; Arnold–Chiari Malformation; Child

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*Corresponding Author: Dae–Yeol Lee <https://orcid.org/0000-0002-5733-2669>

Tel: +82-63-250-1469, Fax: +82-63-250-1464, E-mail: leedy@jbnu.ac.kr

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INTRODUCTION

An Arnold-Chiari type 1 malformation is a congenital or acquired disease of the central nervous system that manifests with various neurological symptoms. Clinically, the malformation represents a displacement of the cerebellum that results in tonsillar herniation below the level of the foramen magnum, thereby disturbing cerebrospinal fluid outflow. Among patients with this malformation, 14%–30% of patients are asymptomatic until adolescence or adulthood.¹⁾ The onset of symptoms can be spontaneous or result from trauma. The prevalence of congenital Arnold-Chiari type 1 malformation is approximately 1 per 1,000 births, but is likely much higher.²⁾ There have been some reported cases of Arnold-Chiari type 1 malformations with precocious puberty, but most of these cases are in boys with an age of onset above 7 years.^{3–5)} Here, we describe the case of the youngest girl experiencing an Arnold-Chiari type 1 malformation as well as precocious puberty, who presented with sudden breast engorgement.

CASE REPORT

A girl, aged 4.5 years, presented with a 6-month history of breast budding and intermittent nipple pain. Her parents noted that she had recently begun to grow more rapidly. She was born as a full-term baby. Her medical history indicated that she was treated for hypoglycemia and respiratory distress syndrome at birth in a neonatal intensive care unit. Her family history demonstrated that her father's and mother's heights were above the 75th percentile on the Korean growth chart. Her mother's menarche age was unremarkable.

The patient's height was 107.3 cm (within the 90th to 95th percentile), and her weight was 18.7 kg (within the 90th percentile). Physical examination and neurological test results were normal. She did not exhibit any neurological signs or symptoms. Her pubertal stages, according to Tanner, were B2 of the breast and P1 of the pubarche (i.e., no axillary hair). The patient's bone age was 6.8 years, which was determined using Greulich and Pyle's method. The gonadotropin-releasing hormone (GnRH) test showed a pubertal response (luteinizing hormone level peak of 13.4 mIU/mL and basal level of 0.6 mIU/mL; follicle stimulating hormone level peak of 12.2 mIU/mL and basal level of 3.6 mIU/mL; estradiol level of 9.29 pg/mL). Thyroid and adrenal functions were normal.

Since puberty began early and was accelerated in the patient, we performed brain magnetic resonance imaging (MRI). Brain MRI showed a pineal cyst and Chiari type 1 malformation (i.e., downward displacement of the cerebellar tonsil by 6 mm). Additionally, no hypothalamic hypophyseal lesions were observed (Figure 1).

Following diagnosis, she was treated with GnRH analogue therapy and leuprolide acetate depot treatment. After 6 months of treatment, she was asymptomatic, her breast budding was slightly improved, and her growth rate became 2.5 cm for 6 months. Furthermore, no neurological symptoms were observed.

DISCUSSION

Congenital Arnold-Chiari type 1 malformation is defined as a tonsillar herniation of 3–5 mm or more;¹⁾ MRI is the imaging modality used most often to diagnose this malformation. The causes for central precocious puberty (CPP) include hypothalamic hamartomas, postencephalitic scars, tubercular meningitis, head trauma, hydrocephalus, tuberous sclerosis, porencephaly, and suprasellar space-occupying lesions, including arachnoid cysts.⁶⁾ Additionally, patients with Arnold-Chiari malformations with myelomeningocele have been reported in some studies.^{7,8)} However, reports concerning patients with Chiari type 1 malformations and precocious puberty are rare. In 1996, Turjman et al.⁵⁾ reported two such cases; however, both these patients had a hypothalamic hamartoma. The concomitant manifestation of Chiari type 1 malformations and precocious puberty was first described by Interlici et al.⁹⁾ in 2000. In that case report, the patient, a boy aged 10.9 years, was treated with neurosurgery, which did not normalize pubertal progression. Since then, Chiari type 1 malformation and precocious puberty have been reported in 3 boys (8.8 years, 9 years, and 10.4 years)³⁾ and 2 girls (7.5 years and 9.5 years).⁴⁾

The posterior cranial fossa of our patient, who exhibited Chiari type 1 malformation, was relatively small for the hindbrain, thereby resulting in inferior tonsillar displacement that induced blockage of cerebrospinal fluid at the foramen magnum between the intracranial and spinal subarachnoid space. The development of Chiari type 1 malformation has been hypothesized to be associated with precocious puberty as the increase in peri/postnatal intracranial pressure caused by impaired cerebrospinal fluid circulation or compression and distortion of the hypothalamus can lead to premature activation of the hypothalamic-pituitary-gonadal axis; however, this association is not yet completely understood.^{3,4)} Nevertheless, the association between Ar-

Figure 1. Sagittal T1-weighted magnetic resonance image of the brain. A pineal cyst (round) and Arnold-Chiari type 1 malformation (arrow; i.e., downward displacement of the cerebellar tonsil of about 6 mm) were observed.

nold-Chiari type 1 malformation and precocious puberty in reported studies can be considered meaningful.

The symptoms and signs of Arnold-Chiari type 1 malformations are relevant to the regions of the brain that are affected, such as the brainstem, cranial nerves, cerebellar cord, or spinal cord.³⁾ Common symptoms include headache, fatigue, muscle weakness, difficulty swallowing, and choking and vomiting, while severe cases result in paralysis.¹⁰⁾ Treatment is still debatable, but decompressive surgery or the management of symptoms is recommended in symptomatic patients with Arnold-Chiari type 1 malformation.⁹⁾

In conclusion, Arnold-Chiari type 1 malformation can be a possible cause of CPP without neurological symptoms in children below the age of 5 years. Furthermore, brain MRI is a good diagnostic tool for evaluating reasonable causes of Arnold-Chiari type 1 malformation in the brain, especially in young patients with precocious puberty.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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