ACFS ARCHIVES OF CRANIOFACIAL SURGERY
Arch Craniofac Surg Vol.14 No.2, 115-118
http://dx.doi.org/10.7181/acfs.2013.14.2.115

남매간에 발생한 두개골조기유합증

최정환[1] · 김명훈[1] · 김민수[1] · 정규용[2] · 이근철[1] · 김석권[1]

[1]동아대학교 의과대학 성형외과학교실, [2]정앤정 성형외과

Craniosynostosis Occurring between Siblings

Jeong-Hwan Choi[1], Myung-Hoon Kim[1], Min-Su Kim[1], Gyu-Yong Jung[2], Keun-Cheol Lee[1], Seok-Kwun Kim[1]

[1]*Department of Plastic and Reconstructive Surgery, Dong-A University School of Medicine, Busan;*
[2]*Jung and Jung Plastic Surgery Clinic, Ulsan, Korea*

Craniosynostosis is a congenital anomaly in which cranial sutures close prematurely and restrict skull growth. In this paper, the case of two siblings, a male and a female, who were both diagnosed as craniosynostosis is reported. They underwent corrective osteotomy for cranial vault remodeling. A 22-month-old female infant who was brought to the department of plastic and reconstructive surgery of the authors' hospital was diagnosed with plagiocephaly. At the same time, her 7-month-old brother was diagnosed with brachycephaly. In the case of the female infant, corrective coronal osteotomy and supraorbital bar advancement were performed. Her brother underwent frontal advancement osteotomy using Tessier's tongue in the groove procedure. After the correction of the craniosynostosis, the two patients recovered in several days later, and the results were good in both cases cosmetically and functionally. They showed normal head circumference increasing curves and no symptom of functional disorder in their last follow-up. Isolated or nonsyndromic craniosynostosis is sporadic but mostly autosomal dominant. This paper presents a case of craniosynostosis with a genetic tendency; and although it occurred between siblings, the affected lesions differed. Thus, appropriate diagnosis and management in patients are needed.

Keywords: Craniosynostosis / Genetic / Siblings

Case Report

Introduction

Craniosynostosis is the premature fusion of one or more of the cranial sutures that can occur as part of a syndrome of congenital anomaly or as an isolated defect called a nonsyndromic. Craniosynostosis occurs in one per 2,000 to 4,000 births, and syndromic craniosynostosis with malformations of other parts of the midface, lower face, axial skeleton, hand and feet occurs in less than 20% of all cases [1]. In some cases, nonsyndromic craniosynostosis occurs among family members. This paper reports a case of craniosynostosis between siblings, one with plagiocephaly and the other with brachycephaly.

Case Report

A 22-month-old girl and her 7-month-old brother, both of whom had irregularly shaped heads, were brought to the author's hospital clinic. There were normal birth histories and no mutations on genetic testing when they were born. Also, there were unremarkable family medical histories and no other craniosynostosis in family and relatives. And they had no multiple craniofacial dysostosis and limb derformities. However, the left side of the forehead of

Correspondence: Seok-Kwun Kim
Department of Plastic and Reconstructive Surgery, Dong-A University School of Medicine, 26 Daesingongwon-ro, Seo-gu, Busan 602-715, Korea
Tel: +82-51-240-2807 / Fax: +82-51-243-5416 / E-mail: sgkim1@dau.ac.kr

*This study was presented at the 69th Congress of the Korean Society of Plastic and Reconstructive Surgeons.

Received August 30, 2013 / Revised October 1, 2013 / Accepted October 7, 2013

the female infant was flat with an elevated superiolateral aspect of orbit that was higher than the right orbital level with right pseudoblepharoptosis. And her brother had a short head as a result of a shortened sagittal axis with a transversely widened axis and supraorbital rim recession on both sides. Through physical examinations, they were suspected as having craniosynostosis or positional plagiocephaly, so computed tomography (CT) with plain radiography was performed to confirm such diagnoses. The radiology results for the female infant showed unilateral left coronal synostosis that was diagnosed with plagiocephaly, and for her brother, bicoronal synostosis with mild metopic synostosis that was diagnosed with brachycephaly. These siblings belonged to the nonsyndromic craniosynostosis type because they had no malformations other than craniosynostosis. They received surgical correction as soon as their craniosynostosis was confirmed. The female infant underwent coronal osteotomy and left supraorbital bar advancement, and her brother underwent frontal advancement osteotomy using Tessier's tongue in the groove procedure and trigonocephaly correction. Both operations resulted in a 1.5 cm advancement length of left side in sister, 1.5 cm advancement length in brother. Several days after the surgical correction of the craniosynostosis, the siblings recovered and their results were satisfactory cosmetically and functionally, without postoperative complications. They showed almost normal head circumference increasing curves and no symptom of functional disorder after about 3 years (Figs. 1, 2). They are still under outpatient observation, but there have been no unusual findings since the procedure.

Discussion

Craniosynostosis may be classified according to the number of involved sutures (simple or complex), the cause of their premature closure (primary or secondary), and whether or not they coexisted with other anomalies (isolated or syndromic). In most cases, cran-

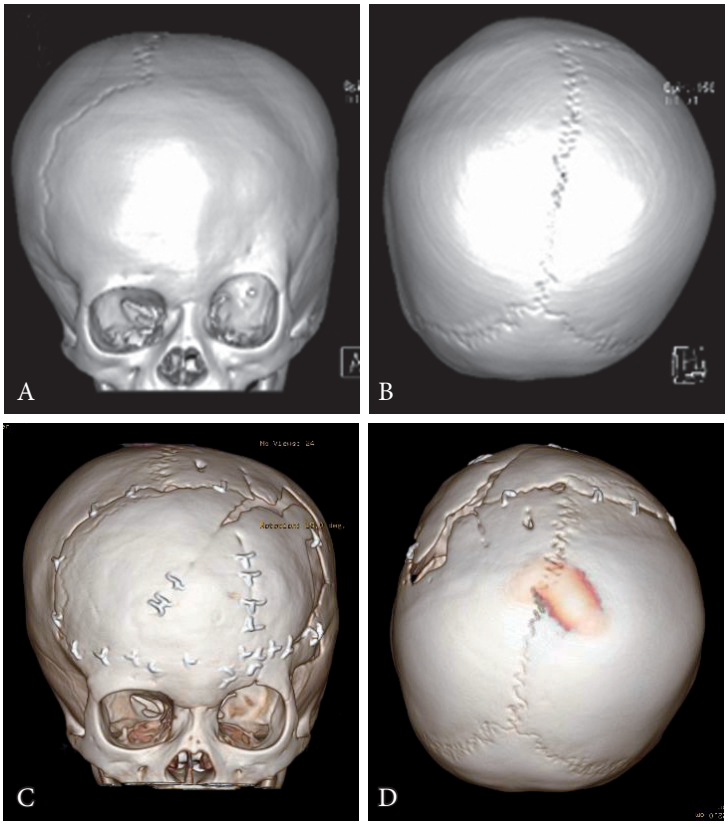

Fig. 1. Preoperative and postoperative three-dimensional computed tomography of the 22-month-old sister. (A, B) Unilateral left coronal synostosis was diagnosed with plagiocephaly. (C, D) Two years after surgical correction.

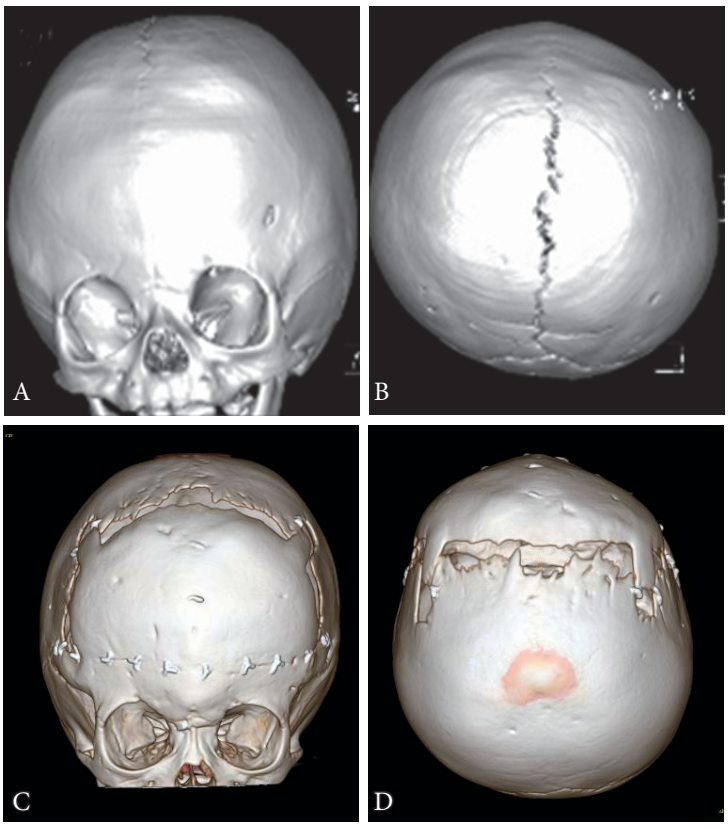

Fig. 2. Preoperative and postoperative three-dimensional computed tomography of the 7-month-old brother. (A, B) Bicoronal synostosis was diagnosed with brachycephaly. (C, D) Two years after surgical correction.

iosynostosis is nonsyndromic, involves a single suture, and has no other malformation. The sagittal suture is affected in 40% to 60% of cases; the coronal suture, in 20% to 30%; and the metopic suture, in less than 10%; and true lambdoid synostosis, which is similar to positional (deformational) plagiocephaly, is rare [1]. The etiology of nonsyndromic craniosynostosis is poorly understood, and the condition is sporadic in most instances. The suspected risk factors are maternal age of 35 years or older, multiple births, a male infant sex, maternal smoking, maternal residence at a high altitude, certain paternal occupations, and fertility treatments [1,2]. Mutation of the fibroblast growth factor and the fibroblast growth factor receptor that regulates fetal osteogenic growth is also involved, which causes Pfeiffer's disease (fibroblast growth factor receptor [FGFR] 1 & 2), Jackson-Weiss syndrome, Apert's syndrome and Crouzon's disease (FGFR2) [2]. Craniosynostosis increases the intracranial pressure and compression of the brain. Untreated progressive craniosynostosis restricts skull growth and causes hydrocephalus, reduced intelligence, developmental delay, visual impairment and an abnormal appearance. Therefore, its early diagnosis and surgical correction is recommended, and its diagnosis relies on physical examination, plain skull radiography and CT. Although there are many types and causes of craniosynostosis, it always requires primary surgical correction. Moreover, the golden time of surgical treatment is controversial, but between 3 and 9 months within 12 months is recommended because approximately more than half of the skull grows within this period [3]. Thus, early and appropriate diagnosis of craniosynostosis is really necessary. When craniosynostosis is suspected, physical examination, including inspection of the craniofacial shape, palpation of the fused suture, and measurement of the head circumference, should be carried out with careful evaluation of the face, neck, spine, digits and toes, because syndromic craniosynostosis can be accompanied by multiple anomalies such as a flat face, malformation of the axial skeleton, a beaked nose, and webbed or fused fingers or toes. Among several

radiographic studies, CT and further three-dimensional (3D)-CT imaging are the standard methods and are essential for the diagnosis of craniosynostosis to recognize the type of premature fusion of the cranial sutures and to plan the surgical intervention [4].

Approximately 39% of infants with all types of craniosynostosis have family history. Among infants with nonsyndromic craniosynostosis, while only 2% of sagittal synostosis patients have a family history of it and others types are even rarer, 8% to 10% of coronal synostosis patients have a family history of it. So coronal synostosis shows a more genetic tendency than any other type of nonsyndromic craniosynostosis [5]. A family study of craniosynostosis at a hospital for sick children in London reported that 12 bicoronal synostosis patients had 9 siblings, one of whom had unicoronal synostosis [6].

The case presented herein involves plagiocephaly and brachycephaly between siblings due to coronal synostosis. They have a high genetic similarity and coronal synostosis phenotypes have the strongest genetic tendency among the different types of craniosynostosis. In this case and in all other cases of craniosynostosis, appropriate and accurate diagnosis and management are really needed. The authors have treated a case of familial nonsyndromic craniosynostosis, and have achieved satisfactory results using the accurate approach and surgical correction.

Although no specific gene mutations in nonsyndromic craniosynostosis have been identified, in several cases, familial nonsyndromic craniosynostosis was found to have a genetic tendency even if the family members have different phenotypes. Thus, appropriate clinical examination of the family members of the involved patient is needed and early diagnosis with surgical management is very important for its prognosis.

References

1. Ferreira MP, Collares MV, Ferreira NP, Kraemer JL, Pereira Filho Ade A, Pereira Filho Gde A. Early surgical treatment of nonsyndromic craniosynostosis. Surg Neurol 2006;65 Suppl 1:S1:S22-6.
2. Kabbani H, Raghuveer TS. Craniosynostosis. Am Fam Physician 2004;69:2863-70.
3. Ursitti F, Fadda T, Papetti L, Pagnoni M, Nicita F, Iannetti G, Spalice A. Evaluation and management of nonsyndromic craniosynostosis. Acta Paediatr 2011;100:1185-94.
4. Cohen SR, Pryor L, Mittermiller PA, Meltzer HS, Levy ML, Broder KW, Ozgur BM. Nonsyndromic craniosynostosis: current treatment options. Plast Surg Nurs 2008;28:79-91.
5. Kimonis V, Gold JA, Hoffman TL, Panchal J, Boyadjiev SA. Genetics of craniosynostosis. Semin Pediatr Neurol 2007;14:150-61.
6. Carter CO, Till K, Fraser V, Coffey R. A family study of craniosynostosis, with probable recognition of a distinct syndrome. J Med Genet 1982;19:280-5.