

Multi-directional Cranial Distraction Osteogenesis for Treating Sagittal Synostosis with Frontometaphyseal Dysplasia: A Case Report

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Summary: Frontometaphyseal dysplasia (FMD), also known as Gorlin-Cohen syndrome, is a rare genetic syndrome. This syndrome affects the skeletal system and connective tissue, and causes a wide spectrum of manifestations of the skull, tubular bones, cardiovascular system, urinary system, and/or gastrointestinal system. Craniofacial findings of FMD are characterized by protruding supraorbital ridge, broad nasal bridge, hypertelorism, down-slanting palpebral fissures, and/or micrognathia. We describe a case of a 2-year-old girl diagnosed with sagittal synostosis accompanied with FMD. She presents anterior sagittal synostosis cranial form, compressed cerebrospinal fluid space (which suggested increased intracranial pressure), and the supraorbital hyperostosis. She underwent multi-directional cranial distraction osteogenesis in the calvaria and shaving of the supraorbital ridges. Despite concerns about bone fragility associated with FMD, the surgery was accomplished as usual. The patient had no intra- and postoperative complications. After 6 months of follow-up, the cranial shape has improved and the cerebrospinal fluid space has widened, but the supraorbital ridge has protruded again. Re-protrusion of the supraorbital ridge appears to be due to age-appropriate vigorous osteogenesis. The multi-directional cranial distraction osteogenesis procedure has been useful for treating sagittal synostosis even concomitant with FMD. (*Plast Reconstr Surg Glob Open* 2021;9:e3551; doi: 10.1097/GOX.0000000000003551; Published online 23 April 2021.)

Frontometaphyseal dysplasia (FMD), reported by Gorlin and Cohen¹, is a type of osteodysplasia caused by FLNA gene mutation and characterized by cranial and tubular bone dysplasia, sometimes accompanied by deafness, progressive joint contracture, and/or internal organ malformations. Its craniofacial findings are characterized by prominent supraorbital ridge, hypertelorism, down-slanting palpebral fissures, and/or micrognathia, occasionally complicated with craniosynostosis. Treatment involves the use of hearing aids for deafness, and surgery for each deformity. We report a case of sagittal synostosis (SS) accompanied with FMD in which the patient underwent cranial distraction osteogenesis.

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CASE REPORT

Preoperative Findings

The patient was a girl noted for deafness during neonatal screening. She exhibited the supraorbital hyperostosis and osteodysplasia of the extremities at 1 year, and clinically diagnosed with FMD. At 2 years and 4 months, she presented with delayed closure of the anterior fontanel, and a computed tomographic scan showed SS, copper beaten appearance, and compressed cerebrospinal fluid spaces (Fig. 1). (See figure, Supplemental Digital Content 1, which shows preoperative patient views that represent the prominent supraorbital ridge, hypertelorism, and mild micrognathia (a); osteotomy line and the position of each pin (b); distractors mounted on a helmet-type frame and connected to the traction pins with steel wire (c, d). <http://links.lww.com/PRSGO/B644>.) She had no developmental delay, but was suggested to have intracranial hypertension (ICH), and then referred to our department for improvement of cranial deformity.

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Surgical Procedure

At 2 years and 8 months, the multi-directional cranial distraction osteogenesis (MCDO) procedure was performed as described previously.^{2,3,4} Briefly, a coronal incision was made, and the entire cranium was divided into 9 bone flaps with a bone saw and an ultrasonic bone scalpel (Sonopet; Stryker, Kalamazoo, Mich.). The supraorbital hyperostosis was shaved with a burr. Traction pins were fixed in each bone flap. After closure of the wounds, a helmet-type frame was fixed by anchor pins in the temporal bones. The wires were fixed as connected the traction pins with the frame-mounted distractors (SDC1). The operative time was 318 minutes. Intraoperative blood transfusion volume was 42 mL/kg body weight. Six days after surgery, distraction was initiated at a rate of 1.5 mm/day. The traction rate was later modified depending on the shape of the cranium. The distraction device was fixed on postoperative day 14, and the device was removed on postoperative day 49. The intracranial volumes before and 6 months after surgery were calculated using the software for 3D reconstruction and analysis (InVesalius 3; CenPRA, Brazil).

RESULT

There were no complications within the follow-up period. At six months postoperative (Fig. 2), the intracranial volume was 1348 mL, increased from 1083 mL preoperatively. Mid-sagittal vector analysis, which was performed according to previously reported methods,⁵ showed increased height of vertex, so that the cranium shape was improved. Cephalic index was increased mildly from 78.6 (Preoperative) to 82.8. The supraorbital ridge

was contoured well at 1.5 months postoperative, but has protruded again 6 months later.

DISCUSSION

FMD is classified as one of the oto-palate-digital spectrum disorders (OPDS) caused by mutations in the gene *FLNA*, which regulates the cytoskeletal protein Filamin A. OPDS present with craniofacial osteodysplasia, cleft palate, deafness, tubular bone dysplasia, and/or the internal organs malformations. *FLNA* is present on the X chromosome (Xq28), and OPDS is an X-linked dominant inheritance. Therefore, the phenotype of female cases varied from mild to severe depending on the degree of skewing of X chromosome inactivation.⁶ As this patient rejected the genetical diagnosis testing, she was clinically diagnosed with FMD because of the supraorbital hyperostosis, the mandible and occipital spur, deafness, micrognathia, brachytelephalangy, the bowing long tubular bones, twisted ribbon rib, and so on.

It has been reported that OPDS and craniosynostosis can be presented by *FLNA* abnormality alone.⁷ In this case, SS is an anterior type with retrocoronal band^{8,9} and ICH was predicted due to enlarged anterior fontanel and copper beaten appearance; therefore, the surgical treatment was recommended. The surgical methods of SS are strip craniectomy, 1-stage cranioplasty, and cranial distraction osteogenesis. We previously described the MCDO procedure.^{2,3,4} Recently in the MCDO procedure, the setting of the osteotomy line is divided into 9 flaps: the anterior-posterior direction is divided into 3 rows of anterior, middle, and posterior segments, and the medio-lateral direction is divided into 3 columns of right, middle, and left segments.



Fig. 1. Preoperative three-dimensional computed tomography imaging. A. Frontal view. B. Lateral view.

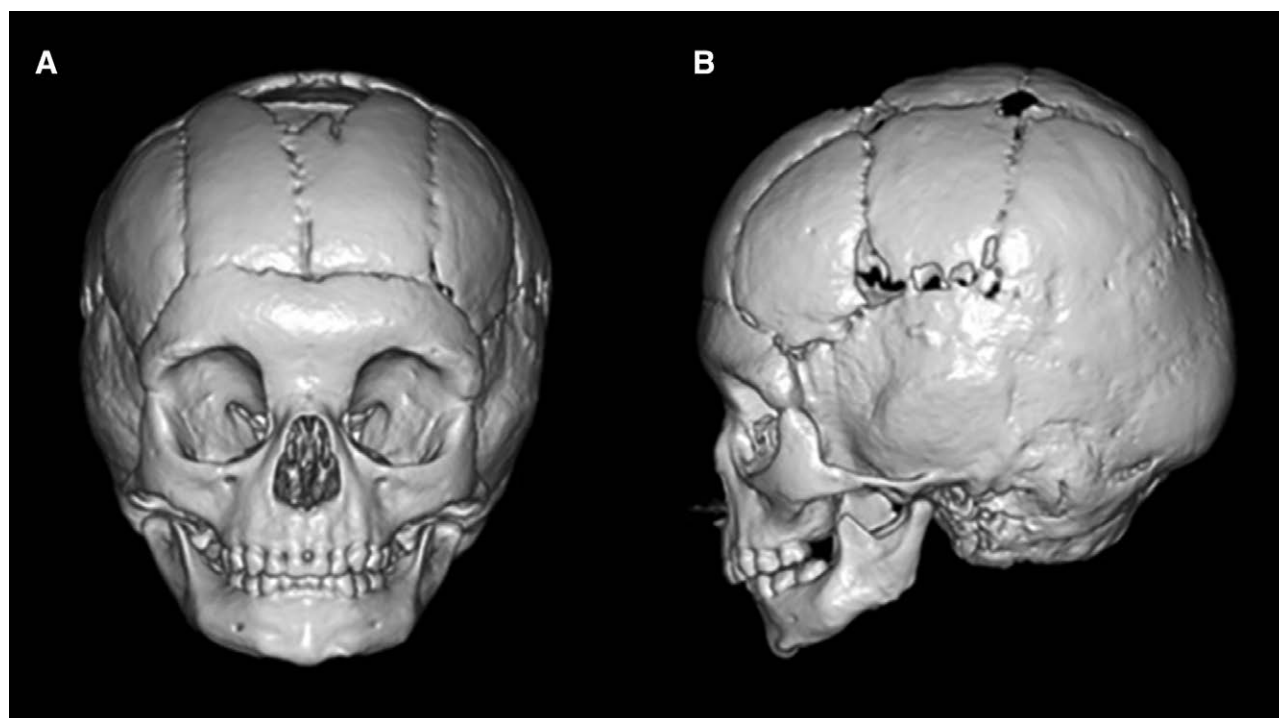


Fig. 2. Three-dimensional computed tomography imaging 6 months postoperative. A. Frontal view. B. Lateral view.

In this case, only the cortical osteotomy was used between the anterior and central segments in the central row to avoid any steps, and the two segments were almost entirely in 1 flap. The position of the traction pins is determined to achieve a good cranial shape. Essentially, 2 traction pins are fixed to each bone flap, so that if one traction pin falls off, the other can be used for traction. In some cases, the bone flap may be left without traction pins so that it can be pulled naturally with the extension of the surrounding bone flap. The direction of traction is perpendicular to the bone flap. Compared with other cranioplasty procedures, the MCDO method allows for vertical bone extension by using a helmet-type frame.

MCDO can achieve both volume expansion and morphological improvement at the same time. Regarding expansion, MCDO is more efficient than 1-stage cranioplasty because it allows for 3-dimensional bone extension. We have previously reported that MCDO requires only 1 cm of cranial expansion compared with 2 cm of one-way bone extension³. We also preserve the periosteum as much as possible and do not strip the bone fragments from the dura, thereby preserving blood flow to the fragments. These 2 advantages lead to better osteogenesis and cranial remodeling. The morphology is easily adjustable in all directions, depending on the position of the osteotomy, the number of fragments, and the degree of bone extension by actually observing the patient's appearance.

Our preoperative concern was the possibility of calvaria fragility due to osteosclerosis with FMD. However, osteotomy was not different from usual, and postoperative bone healing was normal. The prominent supraorbital ridge was corrected appropriately in the surgery. Although

the removed supraorbital ridge prominence has not re-protruded in a case report of adult FMD,¹⁰ our case has been recurrent. One reason for this is probably that the surgery was performed at an age of vigorous osteogenesis. In addition, another reason could be that the amount of bone shaving was insufficient. In adult FMD case reports,¹⁰ the bony cortex is noticeably thickened at the bony protrusion. Therefore, the fact that the protrusion was shaved but the bony cortex was left at least as much as normal may lead to its re-protrusion 6 months after surgery.

We perform the operation before the development of the cranial cancellous bone to minimize the bleeding, and do the same when the patient's osteogenesis is in full swing because the MCDO technique is a type of distraction osteogenesis. For these reasons, we perform the MCDO between the age of 1 and 2 years, but if the patient is referred to our department after that age, or if symptoms of ICH occur later, we perform the surgery as early as possible.

CONCLUSIONS

We performed MCDO for treating SS accompanied with FMD and have a satisfactory result without substantial complications. We have learned from this case that MCDO is a suitable treatment for SS even with osteodysplasia such as FMD. We will continue to perform MCDO for such cases in the same manner, and will do additional osteotomy for the protruding bone with FMD when the patient becomes an adult.

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