Chin-on-Chest Deformity in Patients with Fibrodysplasia Ossificans Progressiva

A Case Series

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Fibrodysplasia ossificans progressiva is a rare and disabling autosomal dominant disorder of joint malformations and progressive heterotopic ossification¹⁻⁴ caused by a recurrent missense point mutation in activin receptor 1A (ACVR1, also called ALK2), a bone morphogenetic protein (BMP) type-I receptor^{2,3,5}. Joints become permanently ankylosed in positions that drastically impair activities of daily living and even life itself⁶. Progressive heterotopic ossification usually begins in the first decade of life, typically appearing first in the dorsal, axial, cranial, and proximal regions of the body, and then later in the ventral, appendicular, caudal, and distal regions^{1-4,7}. Lesions are characterized by the rapid appearance of painful, highly vascular fibroproliferative swellings that progress through an endochondral process to form mature ossicles of heterotopic bone^{3,8}. Lesions may develop spontaneously, although falls⁹, soft-tissue injury^{2,3,10}, mandibular blocks¹¹, intramuscular immunizations¹², viral illnesses¹³, and surgical trauma¹⁰ can precipitate new flare-ups resulting in heterotopic ossification.

Although the classic clinical features of fibrodysplasia ossificans progressiva have been well described, manifestations of the disease affecting the cervical spine are less well known. Neck stiffness is an early finding in most patients and can precede the onset of heterotopic ossification^{2,4,10}. Characteristic malformations of the cervical spine include large posterior elements; tall, narrow vertebral bodies; and fusion of the facet joints from C2 to C7^{2,14,15}. In the present report, we describe a rare, but potentially life-threatening, chin-on-chest deformity that may develop in patients with fibrodysplasia ossificans progressiva and a surgical strategy that has been effective for achieving substantial correction of the deformity. The families were informed that data concerning these cases would be submitted for publication and they consented.

Case Reports

CASE 1. A nine-month-old girl had severe manifestations of fibrodysplasia ossificans progressiva involving the head and neck, resulting in a severe right-sided torticollis from involvement of the sternocleidomastoid muscle. When we first saw the patient at the age of sixteen months, she also had a kyphosis of the lower cervical spine. Both deformities were rigid and progressive. A three-dimensional reconstructed computed tomography scan (Fig. 1-A) allowed us to hypothesize that the deformity in the cervical spine may have started superiorly, between the occiput and C2 with a rotatory subluxation. Subsequently, fusion at the level of the right C2-C3 facet joint occurred, resulting in asymmetric occipitalization and a torque on the neck that resulted in the chin-on-chest deformity (Fig. 1-B).

By the age of thirty months, the patient had restrictive chest wall disease¹⁶, which was further complicated by influenza A pneumonia and respiratory arrest. She was resuscitated and intubated, while awake, through a fiber-optic nasotracheal approach¹⁷⁻¹⁹. She experienced prolonged, severe acidosis and remained intubated for one month. Efforts to wean her from the ventilator were unsuccessful.

One month later, she was transferred to our institution for combined orthopaedic and otorhinolaryngologic evaluation and treatment. The chin-on-chest deformity was severe, and a tracheotomy could not be performed safely because of the severe deformity of the neck, which completely prevented access to the trachea (Fig. 1-B).

The patient was managed with glucocorticoids (prednisone [2 mg/kg once daily], administered intraoperatively and for two days postoperatively) to abrogate severe reactive soft-tissue swelling that occurs following soft-tissue trauma in patients with fibrodysplasia ossificans progressiva²⁰. A multiple-

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Fig. 1-A

Fig. 1-B

Fig. 1-C

Fig. 1-D

Figs. 1-A through 1-D Case 1. **Fig. 1-A** Computed tomography scan with three-dimensional reconstruction, made at the age of twenty months (ten months before surgery), demonstrating torticollis and flexion deformities. **Fig. 1-B** Lateral radiograph, made at the age of thirty months (before operative correction), demonstrating a rigid chin-on-chest deformity. **Fig. 1-C** Lateral radiograph made one month after surgery, demonstrating that the chin has been extended to a neutral position and is being stabilized by a halo vest. The tracheostomy can be visualized in the anterior part of the neck. **Fig. 1-D** Photograph made two years after surgery and removal of the halo vest, showing the neck in a neutral position. At the time of the latest follow-up, five years after surgery, the neck remained in a stable position.

Fig. 2-A

Fig. 2-B

Fig. 2-C

Figs. 2-A, 2-B, and 2-C Case 2. Lateral radiograph (Fig. 2-A), sagittal T1-weighted magnetic resonance image (Fig. 2-B), and clinical photograph (Fig. 2-C), made at the age of fifteen years, demonstrating a severe, rigid, chin-on-chest deformity.

pin halo ring was applied with the patient in a prone position on a well-padded operating room table. A posterior cervical exposure and Smith-Petersen osteotomies were performed to the nerve roots from C1-C2 to C7-T1²¹. Following the osteotomies, there was sufficient spinal flexibility to allow manipulation. Allograft and autograft bone was applied over the decorticated lateral masses and facet joints, and a halo vest was applied to maintain reduction in the corrected position. With adequate anterior exposure obtained, a tracheotomy was performed without difficulty (Fig. 1-C).

The patient was transferred to a local hospital in her home state one month after surgery. The halo vest was removed

thirteen weeks postoperatively, and ventilator weaning began; however, the patient remained ventilator-dependent. Five years after surgery, the neck was stable and in an excellent position, with a solid posterior fusion from C1 to T1 (Fig. 1-D).

CASE 2. A seven-year-old girl had development of heterotopic ossification involving both shoulder girdles. Subsequently, she had stiffness of the neck, increasing thoracic and lumbar scoliosis, and swelling of the anterior strap muscles of the neck, with a subsequent and rapid decrease in the range of motion of the neck. The patient was examined by us at the age of eight years, at which time the range of motion of the cervical spine was limited to 20° of flexion, 0° of extension, 15° of left

Fig. 3-A

Fig. 3-B

Figs. 3-A and 3-B Case 3. **Fig. 3-A** Photograph made before surgery, demonstrating a severely disabling, rigid, chin-on-chest deformity. **Fig. 3-B** Photograph made twenty years after surgery, demonstrating that lateral tilt of the neck has occurred since the original operative correction; however, the neck has remained extended in a nearly neutral position.

lateral bending, 0° of right lateral bending, and 15° of rotation bilaterally. Hyperemia without tenderness was noted along the anterior strap muscles and right sternocleidomastoid muscle. Radiographs demonstrated characteristic malformations in the cervical vertebrae and congenital fusion of C5-C6 but no other unusual features.

The patient was next seen eight years later, at the age of fifteen years, after the development of a severe chin-on-chest deformity (Figs. 2-A, 2-B, and 2-C). Fibrodysplasia ossificans progressiva involving the sternocleidomastoid muscle created an osseous bar that tethered the head and face to the sternum and pectoral region, resulting in a fixed deformity, even as the spine continued to grow. Operative correction of the deformity could not be offered because adequate exposure could not be achieved safely as a result of both anterior blockage (caused by the mandible, which was firmly pressed to the chest) and posterior blockage (caused by the shoulder as well as by the deformity itself). Access to the osseous bar was limited to <1 cm on either side. Because of the location of the neurovascular structures that lie in direct juxtaposition to the osseous bar, we concluded that the risks of resecting the osseous bar, including uncontrollable bleeding, were simply too great to attempt operative correction of the deformity. The patient languished at home in social isolation and remained on a soft diet with difficulty swallowing.

CASE 3. An eight-year-old girl who had been previously asymptomatic noticed a neck deformity that progressed rapidly over the course of the subsequent three years. The patient first noticed a nodule on the right side of the neck and had occasional flare-ups consisting of severe pain and swelling in the neck. As a rigid chin-on-chest deformity developed, the patient complained of difficulty eating and progressive loss of visual fields due to the resulting downward direction of her gaze. After three years of nonoperative treatment consisting of nonsteroidal anti-

inflammatory medications, the patient ultimately sought operative treatment at the age of twelve years (Fig. 3-A).

One week before surgery, the patient was admitted to the hospital and was placed in cervical traction in an attempt to correct the cervical kyphoscoliosis. Following minimal correction, the decision was made to perform an operative correction of the chin-on-chest deformity. Multiple Smith-Petersen osteotomies of the cervical spine were performed, and gentle reduction of the kyphoscoliosis was obtained so that the head could be placed in a neutral position. As in the case of the first patient described above (Case 1), no anterior osteoclasis was required because of the lack of involvement of the intervertebral discs. Only local autogenous bone graft was placed along the osteotomized posterior elements following reduction. To maintain reduction, a halo vest was applied for three months. The patient reported great satisfaction with the correction of the cervical deformity. She was able to attend college with assistance in the activities of daily living. Twenty years after the procedure, she was employed and worked as a tax analyst. Since the time of the operation, lateral displacement of the neck had redeveloped, but the patient reported being satisfied with the results of the procedure (Fig. 3-B).

Discussion

We report the cases of three patients who had severe chin-on-chest deformity as a result of progressive heterotopic ossification. Patients with fibrodysplasia ossificans progressiva have orthotopic fusion of multiple levels of the cervical spine independent of heterotopic ossification, thus limiting the number of motion segments and creating a lever arm for deforming forces¹⁵. Unlike most patients with fibrodysplasia ossificans progressiva, the patients described here had asymmetric lesions of the neck. The combination of decreased motion segments resulting in a lever arm for deforming forces,

along with asymmetric lesions, likely initiated the rapid development of the chin-on-chest deformity. As the neck continues to grow while the chin is rigidly tethered to the chest, a severely disabling deformity may develop, highlighting the need for careful surveillance and early operative intervention in selected patients.

Early detection of an impending chin-on-chest deformity is paramount, and in our opinion the use of a hard collar, or in some cases a halo vest, if tolerated, may prevent progression of these deformities. Once an asymmetric neck lesion is identified, we recommend radiographic surveillance to further characterize the nature of deformity progression and to minimize the risk of the development of ossification in a severely deformed position that requires cervical osteotomies or other surgical intervention.

At the present time, no medical or surgical intervention can alter the natural history of fibrodysplasia ossificans progressiva, although many attempts have been made²⁰. Surgical resection of heterotopic bone is generally contraindicated. Patients are commonly more disabled after surgery than they are before surgery because of the rapid, often explosive, recurrence of heterotopic ossification in response to the operative trauma^{1,2,22}. In a case series in which surgical intervention was used to correct thoracolumbar spinal deformity in patients with fibrodysplasia ossificans progressiva and scoliosis, the outcomes were generally poor because of both progression and recurrence of deformity as well as heterotopic ossification at both the operative site and remote sites²³. When surgery is performed, meticulous attention must be given to padding and positioning of the entire body on the operating room table, and perioperative administration of corticosteroids is recommended to counteract the development of heterotopic ossification²⁰. The risks of anesthesia in patients with fibrodysplasia ossificans progressiva are substantial^{17-19,24}, and traditional teaching has emphasized the avoidance of elective surgical intervention in these patients²⁰. However, favorable surgical outcomes have been described in a few case reports^{25,26}.

In our case series, an operative approach was attempted in two patients with severe and progressive chin-on-chest deformities. Successful posterior cervical fusion was accomplished by means of posterior cervical osteotomies, gentle re-

duction of the deformity, and halo vest immobilization, with use of bone graft and without instrumentation. The timing of surgical intervention is controversial and should be considered on a case-by-case basis. Our general recommendation is to intervene surgically once severe progression of the deformity has been documented but before the spine has ankylosed into a nonfunctional position.

We do not routinely recommend the repositioning of joints in patients with fibrodysplasia ossificans progressiva as these procedures entail substantial risk for disease-worsening and life-threatening complications. However, extraordinary, life-threatening circumstances may prevail as in the case of one of our patients (Case 1) for whom few other viable options remained. The need for close follow-up of patients with fibrodysplasia ossificans progressiva lesions in the neck is highlighted by the rapid progression of severely disabling deformity as described in the present report. It is important to identify patients who are at risk for the development of a severe deformity so that steps can be taken to minimize the deformity before it has progressed beyond the point at which it can be addressed safely. ■

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