Case Report

Hurler syndrome: Etiology, manifestations, and life complications: A case report

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ABSTRACT

Hurler syndrome is a rare genetic lysosomal storage disorder with a wide range of manifestations and complications ranging from musculoskeletal deformities to cardiac and corneal problems. The life expectancy of diagnosed patients does not usually exceed 10 years of age due to the associated cardiac problems. However, depending on the complication presented, some treatment modalities are offered to enhance the quality of life for these patients. Our case report reviews a case of a set of twins diagnosed with the syndrome, who have undergone bilateral coronoidectomy via a coronal approach. This approach was taken due to the superior extension of the coronoid processes into the infratemporal fossae bilaterally, their low zygomatic arches, and the severely limited mouth opening associated with coronoid hyperplasia. A coronal approach that included a bilateral coronoidectomy with the removal of internal exophytic bone on the zygomatic arch, along with manipulation under sedation and further physiotherapy/OraStretch device was used to reach the final maximum mouth opening of 35 mm for both patients.

Key words: Coronal flap, Coronoid hyperplasia, Coronoidectomy, Hurler syndrome, Limited maximal interincisal opening, Trismus

Hurler syndrome is an autosomal recessive disease caused by a faulty gene on chromosome 4 that encodes for the enzyme alpha-L-iduronidase (IUDA) [1]. Although the disorder is not very common, more than 250 cases have now been reported worldwide [2]. This enzyme is in charge of breaking down the glycosaminoglycans (GAGs); dermatan sulfate and heparin sulfate [1]. The aforementioned abnormality eventually leads to the accumulation of immense levels of GAGs in the body, causing the cells to become dysfunctional and ultimately leading to death [1]. Hurler syndrome is the prototype of the mucopolysaccharidoses disorder; referred to as mucopolysaccharidosis 1 (MP1) while the other less severe type is referred to as MP2 or Hunter's syndrome [3]. GAG deposition causes organomegaly and thickening, including the heart, spleen, liver, muscles, connective tissues, joints, and the central nervous system, resulting in significant functional impairment [1]. As for treatment, allogeneic hematopoietic stem cell therapy has shown prompt engraftment of gene-corrected cells along with supraphysiological levels of the IUDA enzyme [4]. This has been proposed as a suggested, partially successful modality with complications [4] such as frontalis muscle weakness, hematoma, and infection [5].

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Our main aim here is to investigate the extraoral approach of coronoidectomy in treating coronoid hyperplasia, which is considered to be one of the skeletal manifestations of this syndrome [6].

CASE PRESENTATION

Our case involves twin male siblings, 12 years old, presenting to the clinic for preoperative evaluation before a bilateral coronoidectomy procedure. Both patients present with a history of Hurler syndrome with coronoid process enlargement jaw cramps and spasms. Both patients have a family history of diabetes and bleeding disorder and were born to a father with arrhythmias and a mother with hypoglycemia. They have another sibling as well who was diagnosed with Hurler syndrome. All three are difficult to intubate during procedures.

Our first patient is a 12-year-old male who presents to the clinic with a past medical history of congenital heart disease, valvulopathies, aortic root dilation, obstructive sleep apnea, enlarged spleen and liver in infancy, cerebrovascular accident/trans ischemic attack with residual deficits, seizure disorder, pseudotumor left ventriculoperitoneal (VP) shunt, hypothyroidism, scoliosis, gait disturbance, corneal clouding, and bilateral coronoid process hyperplasia extending into bilateral

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temporal fossa. Previous surgeries include patent foramen ovale (PFO) closure, pressure-equalizing tubes, VP shunts, spinal fusion, trigger finger- all digits, and double hip osteotomy. Current medications include omnitrope, oxcarbazepine 300 mg oral tablets, 1.5 tablets(s) 300 mg, Vitamin D3: Oral, and 1 tab (s), once a day. The patient was allergic to morphine and oxycodone.

The second twin also presents to the clinic with similar manifestations to his sibling in addition to mild hearing loss in the left ear and growth hormone deficiency. Previous surgeries include PFO closure, ophthalmic strabismus surgery, cervical spinal fusion surgery, trigger finger-all digits, and bilateral hip osteotomy. Current medications are the same as above. The patient is allergic to ceftriaxone, clindamycin, diphenhydramine, latex, diphenhydramine, cefdinir, oxycodone, and morphine.

On examination, both patients presented with a limited maximal interincisal opening (MIO) of about 8 mm due to enlarged coronoid processes and were scheduled for bilateral coronoidectomy (Fig. 1). Bilateral coronoid hyperplasia was confirmed in computed tomography (CT) scans (Figs. 2 and 3). A coronal/bitemporal approach was performed for the coronoidectomy procedure due to the superior extension of the coronoid processes into the infratemporal fossae bilaterally, the limited MIO, and the low zygomatic arches, making proper intraoral access in both patients is not a feasible option.

In our case, both patients underwent general anesthesia and were surgically prepared, draped, and intubated to start the surgical procedure. A bow-like incision drawing was made using a sterile marker slightly anterior to the vertex, leaving about 5 mm of hairline ahead. Both of the twins have previously had VP shunts placed and after a discussion with the neurosurgeon, the decision was made to keep the incision a minimum of 2 cm away from the shunts, which limited the posterior extent of our incision. The drawing extended from one superior temporal line to the other, within the upper regions of the temporal muscle. To minimize intraoperative bleeding, injections of epinephrine 1: 100,000 were given along the entire incision outline. Hair was prepared and an incision was made using no. 10 blade, following the pre-drawn line to the depth of the pericranium. Hair and wound margins were retracted on either side of the incision using hemostatic clips (raney clips), and electrocauterization (Bovie) was used to minimize bleeding during dissection. Dissection in a subgaleal plane was done starting from the superior temporal line, extending into the tempo-parietal fascia, and ending at the zygomatic arch.

Seldon retractor was used to continue the inferolateral extension in a subgaleal plane down to the zygomatic arch and then a #10 blade was used to incise through the skin, subcutaneous tissue, and temporoparietal fascia to avoid incidental damage to the temporalis muscle. Alternatively, Metzenbaum scissors can be used to achieve the same result limiting bleeding from the temporalis muscle. A preauricular extension of the incision to the level of the earlobe was done within a preauricular skin fold and the skin was undermined at the level of temporalis fascia with the preauricular muscles being disconnected. A bilateral dissection was used to elevate the coronal flap anteriorly. Sharp dissection

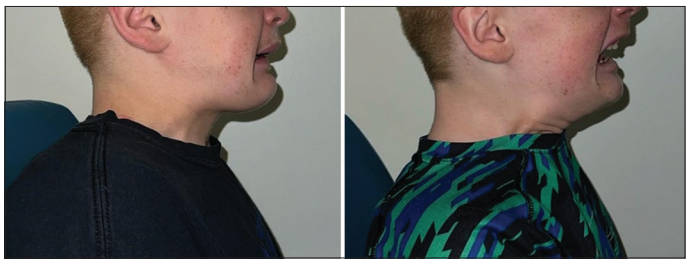

Figure 1: Lateral view showing maximal incisal opening of both twins

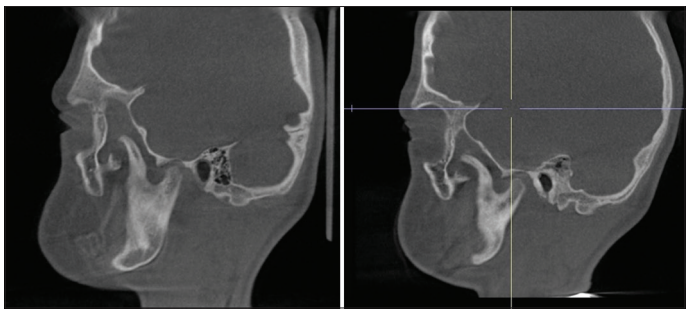

Figure 2: Left and right sagittal computed tomography scans for the first pa-tient showing coronoid hyperplasia

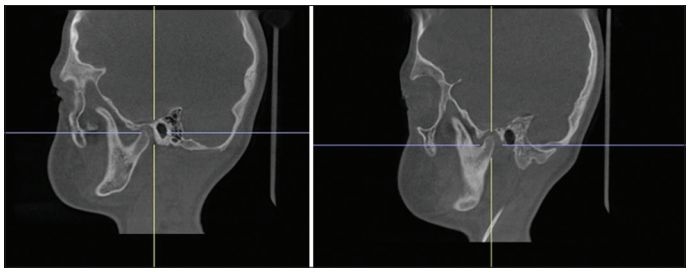

Figure 3: Left and right sagittal computed tomography scans for the second patient showing coronoid hyperplasia

was used to expose the silvery white temporalis fascia over the temporalis muscles (Fig. 4).

Electrocautery was used to expose the frontal and parietal portions of the flap by cutting the loose connective tissue overlying the pericranium. The zygomatic arch along with the coronoid process was exposed. Total coronoidectomy was done on each side with further removal of exophytic bone on the inner portion of the zygomatic arch to reach a good initial mouth opening of about 20 mm. Coronoid cuts and reduction of the inner zygomatic arch were performed using Stryker Sonopet. Alternatively, a reciprocating saw and a round bur could be used. Closure was done in layers, with the placement of a final surgical drain and surgical staples over the incision line. The final injection of epinephrine 1: 100,000 was used to decrease postoperative bleeding as well. Both patients came in about 10 days after for manipulation of their muscles and jaws under general anesthesia, to further increase mouth opening. Surgical staples were also removed during the sedation visit. Both patients were also scheduled for further physiotherapy, later on, and were given instructions on their regular use of the OraStretch device at home to help with their enhanced mouth opening and muscular stretching. Their final maximal incisal opening was recorded to be 35 mm (Fig. 5).

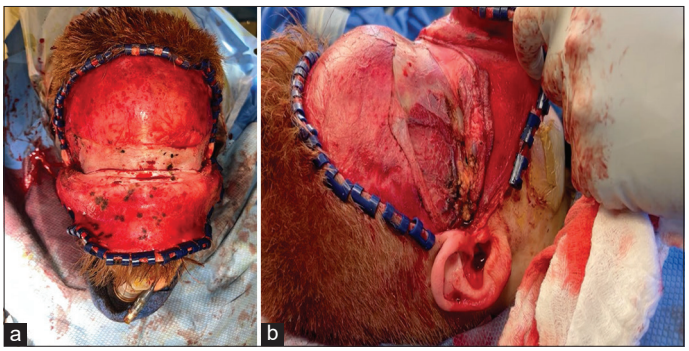

Figure 4: (a) coronal/bitemporal flap reflection; (b) extension of the flap showing the coronoid process on the patient's right side

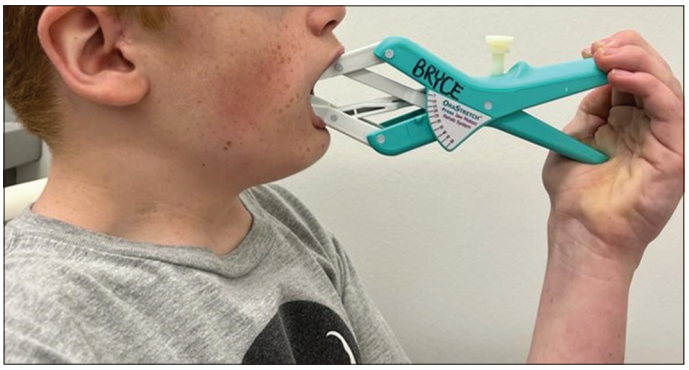

Figure 5: Regular use of the OraStretch device further increased maximal incisal open-ing

DISCUSSION

Our main aim was to highlight how one of the several manifestations of this syndrome was targeted to enhance the patient's quality of life. Dwarfism, severe skeletal deformities, limitation of joint movement, deafness, hepatosplenomegaly, cardiac abnormalities, characteristic facies (being coarse and puffy) [3], and in some cases, clouding of the cornea with eventual opacity and mental retardation are the most common clinical signs of the condition [2]. Oral and dental changes can also be found in these patients, including but not limited to the face, skull, dentition, temporomandibular joint (TMJ), gingiva, the alveolar process, tongue, and palate as well [3]. Manifestations might be as follows: The face: Frontal bossing, supraorbital ridges, temporal bulge, a flattened nasal bridge, and hypertelorism [3]. The skull: Larger than normal with elongated anteroposterior dimension (dolichocephaly) [3]. The dentition: Teeth are smaller than normal and often spaced out [3]. The TMJ: Bilateral concavity or flattening of the articular eminence [3]. Shortening of the condylar neck, enlargement of the coronoid process, and limited joint motion [3].

One cohort study showed that manifestations of Hurler syndrome can appear during the 1st month of age, however, additional studies are needed to establish how these early manifestations can predict how far we can go with treating the syndrome [7]. The mandibular coronoid process, which joins the temporalis muscle and tendon, is removed during coronoidectomy procedures. It is used to treat limited mouth-opening issues caused by submucous fibrosis, TMJ ankylosis, and coronoid process hyperplasia as in our case [8]. A case report by Yura *et al*. highlighted the effectiveness of coronoidectomy in enhancing limited mouth opening in a patient with unilateral coronoid hyperplasia [9]. A different case report by Starch-Jensen and Kjellerup also further highlighted how bilateral coronoid hyperplasia can greatly limit MIO; emphasizing the importance of both coronoidectomy and further vigorous physical therapy to treat the condition [10].

Mandibular coronoid process hyperplasia is a rare congenital or developmental TMJ disorder characterized by coronoid process elongation, which can result in restricted mandibular movement due to interference between the hyperplastic coronoid process and the medial surface of the zygomatic arch [10]. It is associated with temporalis muscle hyperactivity and plays a key role in mandibular elevation with the masseter and medial pterygoid muscles, limiting mandibular movement [8]. Restricted mouth opening has multiple health consequences including difficulty speaking, swallowing, and even compromised oral hygiene [11]. The temporalis muscle is removed from the coronoid process during coronoidectomy, and the jaw is free of temporalis muscle movement [8]. Coronoidectomy and temporalis muscle separation from the mandible have shown excellent clinical efficacy in enhancing mouth opening and movement [8]. Therefore, our aim, in this case, was to increase and enhance mouth opening through a bilateral coronoidectomy procedure. Due to the very limited mouth opening, which was initially measured to be 8 mm, the superior extension of the coronoid processes bilaterally into the infratemporal fossae, and the low zygomatic arches, the usual intraoral approach for this procedure could not be taken. An extra-oral, coronal/bitemporal flap approach was used for better access and visualization. The coronal incision is a widespread and adaptable surgical technique for the anterior cranial vault and upper and middle portion of the facial skeleton [12], where it preserves the integrity of the TMJ when also used for joint-related procedures [13]. The flap itself allows for extensive exposure of the fractures in this area [12]. The size and location of the incision, as well as the layer of dissection, are determined by the surgical procedure and anatomic area of interest [14].

CONCLUSION

Hurler syndrome has a myriad of life-limiting complications. From an oral and maxillofacial surgery point of view, coronoid hyperplasia can result in a severely limited MIO (trismus) which can greatly affect the quality of life and limit the patient to properly take care of their oral hygiene. In cases where the coronoid processes are present superiorly in the infratemporal fossae, the coronal flap approach is a valid technique for better access and visualization when performing coronoidectomy to further enhance their maximal opening.

REFERENCES

1. Sakuru R, Bollu PC. Hurler syndrome. In: StatPearls. Treasure Island, FL: StatPearls Publishing; 2022.

2. Blattner RJ. Hurler's syndrome. J Pediatr 1966;69:313-5.
3. Gardner DG. The oral manifestations of Hurler's syndrome. Oral Surg Oral Med Oral Pathol 1971;32:46-57.
4. Gentner B, Tucci F, Galimberti S, Fumagalli F, De Pellegrin M, Silvani P, *et al*. Hematopoietic stem- and progenitor-cell gene therapy for hurler syndrome. N Engl J Med 2021;385:1929-40.
5. Pogrel MA, Perrott DH, Kaban LB. Bicoronal flap approach to the temporomandibular joints. Int J Oral Maxillofac Surg 1991;20:219-22.
6. Khadembaschi D, Arvier J, Beech N, Dolan D, McGill J. Successful management of severe coronoid process hyperplasia in a patient with mucopolysaccharidosis VI: A case report. J Surg Case Rep 2020;2020:rjz388.
7. Kiely BT, Kohler JL, Coletti HY, Poe MD, Escolar ML. Early disease progression of Hurler syndrome. Orphanet J Rare Dis 2017;12:32.
8. Seok H, Ko SO, Baek JA, Leem DH. Coronoidectomy for reduction of superolateral dislocation of mandible condyle. J Korean Assoc Oral Maxillofac Surg 2022;48:182-7.
9. Yura S, Ohga N, Ooi K, Izumiyama Y. Mandibular coronoid hyperplasia: A case report. Cranio 2009;27:275-9.
10. Starch-Jensen T, Kjellerup AD. Bilateral elongated mandibular coronoid process and restricted mouth opening: A case report. Open Dent J 2017;11:670-8.
11. Gupta H, Tandon P, Kumar D, Sinha VP, Gupta S, Mehra H, *et al*. Role of coronoidectomy in increasing mouth opening. Natl J Maxillofac Surg 2014;5:23-30.
12. Kumar VS, Rao NK, Mohan KR, Krishna L, Prasad BS, Ranganadh N, *et al*. Minimizing complications associated with coronal approach by application of various modifications in surgical technique for treating facial trauma: A prospective study. Natl J Maxillofac Surg 2016;7:21-8.
13. Obwegeser HL. Temporal approach to the TMJ, the orbit, and the retromaxillary-infracranial region. Head Neck Surg 1985;7:185-99.
14. Coronal Approach. [n.d.]. Available from: https://surgeryreference.aofoundation.org/cmf/trauma/midface/approach/coronal-approach [Last accessed on 2023 Aug 11].

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