Surgery first approach for dentofacial deformity correction of a patient with achondroplasia: a case report

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Background: Achondroplasia is a characteristically short physical appearance particular interest to this paper is the craniofacial features. Craniofacial features include a hypoplastic midface, enlarged calvaria, frontal bossing, flattened nasal bridge, mandibular prognathism (potentially masking micrognathia) and dental malocclusion including an anterior open bite.

Case Description: A 19-year-old male with achondroplasia presented to the University of Alabama Birmingham School of Dentistry with the chief complaint that "he needed to eat and chew better". The patient said that in addition to his difficulty incising and chewing foods, he also had speech problems related to jaw position. Clinical examination revealed concave facial profile, hypoplastic midfacial soft tissue, adequate distance between the throat and chin. His malocclusion was complicated by a significant anterior crossbite with −9 mm overjet, left side posterior crossbite, and significant deep overbite. The maxillary and mandibular dental midline was coincident with the facial midline. There was no popping, clicking, or crepitation of the temporomandibular joint. The final treatment plan decided was a surgery first approach (SFA) to rehabilitate the patient. This plan included non-extraction treatment, followed by a Le Fort 1 maxillary osteotomy to advance the maxilla, bilateral sagittal split setback with intermaxillary fixation screws. Post-surgical orthodontic treatment with the use of conventional orthodontic brackets was also planned to correct inter-arch discrepancies after the surgery.

Conclusions: While SFA has been documented for correction of severe dentofacial deformities including cleft lip and palate, severe malocclusions, and hemifacial microsomia, this case report represents the first time a patient with achondroplasia has had a successful SFA to correct his dentofacial deformity and occlusion. This technique represents a breakthrough in the management of patients with significant facial deformities and the effective use of precious financial and clinical resources.

Keywords: Surgery first; achondroplasia; 3D imaging; virtual surgical planning (VSP); case report

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Introduction

Background

Achondroplasia is the most common form of dwarfism in humans. Achondroplasia is a rare condition and occurs in only 1 in 26,000 people (1). In the United States today there are approximately 5,000 patients living with achondroplasia (2). Achondroplasia is an autosomal dominant trait (3). Children who inherit the affected gene type from both parents generally die *in-utero* or early infancy. Achondroplasia often arises from a substitution mutation in the fibroblast growth factor 3 receptor (*FGFR3*) gene on chromosome 4p16.3. Nearly all cases of achondroplasia are the result of a new mutation of this gene. Normal function of *FGFR3* is the inhibition of chondrocyte proliferation (3). Mutation of *FGFR3* gene results in overactivity of the FGFR3 protein and suppressed proliferation and maturation of growth plate chondrocytes (3). Defective endochondral osteogenesis induces premature ossification of the epiphyseal plates of long bones, as well as, the synchondrosis in the cranial base and spine (4). As such, achondroplasia is attributed to a quantitative loss of tissues rather than abnormal tissue formation. The result of this is that patients with achondroplasia exhibit characteristic physical appearance, as well as various systemic complications.

There are many clinical signs and the systemic complications due to achondroplasia are numerous. Perhaps the most obvious sign is a characteristically short physical appearance (median height for males and females is 131 and 124 cm respectively) (5). Craniofacial features include hypoplastic midface, enlarged calvaria, frontal bossing, flattened nasal bridge, mandibular prognathism (potentially masking micrognathia), and dental malocclusion including anterior open bite (3,5-7).

Achondroplasia results in early closing of these synchondroses with the spheno-occipital more affected than the spheno-ethmoidal (4,7). As a result, the anterior cranial base in patients with achondroplasia is approximately normal length; however, the posterior cranial base is severely shortened (4,7). Shortening at the cranial base causes the viscerocranium to remain in a backward position. This lack of forward movement inhibits the downward and forward growth of the maxilla resulting in characteristic midface deficiency. Interestingly, mandibular growth is unaffected because condylar cartilage growth is the product of an unaffected process (periosteal chondrogenesis) (3). As a result of normal mandibular growth and a deficient maxilla, the most common skeletal malocclusion in patients with achondroplasia is Angle Class III (4,5,7). In addition to the dentofacial deformity, the prevalence of obstructive sleep apnea (OSA) in patients with achondroplasia is nearly 40% with symptoms typically presenting from ages 2 to 10 (6).

There is currently no cure for achondroplasia. Management of achondroplasia focuses on maximizing functional capacity while minimizing symptoms and complications. Growth hormone treatment decreases chondrocyte inhibition and has been shown to improve limb length in achondroplasty children. There is limited literature documenting the correction of craniofacial abnormalities due to achondroplasia using orthognathic surgery (5). Correction of skeletal Angle Class III and OSA is accomplished by moving the entire midface forward via a LeFort III osteotomy (4). A 2017 case report illustrated the ability of conventional orthodontic treatment alone in achieving acceptable occlusion, however, the current guideline for management of skeletal malocclusion for patients with achondroplasia applies a three-stage, surgical-orthodontic approach (3,5).

Rationale

The conventional approach to surgical-orthodontic treatment calls for three stages: preoperative orthodontics, surgery and postoperative orthodontics (8). Preoperative

Highlight box

Key findings

- The clinical concept of the "surgery first approach (SFA)" to craniofacial deformities is becoming more popular and is practiced by many clinical teams worldwide. This case report describes a unique case of the "SFA" on a patient with achondroplasia in a setting where the patient's insurance coverage was coming to an end. Unique challenges and the treatment plan are described in this case report.

What is known and what is new?

- Surgery first is not a new concept and has been reported in the literature.
- This manuscript describes the SFA for a patient with achondroplasia during the COVID-19 pandemic.
- The manuscript also describes a very challenging situation where the patient's insurance was running out.

What is the implication, and what should change now?

- This paper describes the efficiency when craniofacial teams co-operate to help the patients' diagnosis, treatment plan and execution of the clinical management.

orthodontic therapy contributes to optimal surgical outcomes by teeth realignment relative to supporting bone, arch coordination and axial correction of the incisors (4,8,9). Over the years, the three-stage method has been validated by producing acceptable levels of stability and patient satisfaction in post-treatment outcomes (8,10,11). However, this approach has its drawbacks including lengthy orthodontic treatment (15–24 months preoperatively; 7–12 months postoperatively).

In recent years, the advancement of 3D imaging technology (12-14), computer-aided design, computer-aided manufacturing (CAD/CAM) technologies (15,16) and improved communications between surgical teams have resulted in a surgery first approach (SFA) in many conventional orthodontic and orthognathic surgical cases (17-19). The current concept of the SFA is a team approach involving surgeons and orthodontists (20). The main advantages of the SFA are reduced treatment duration, improved facial profile from the onset of treatment, high patient and orthodontist satisfaction rates, efficient and effective orthodontic decompensation, rapid patient recovery, and immediate increase in the upper airway dimensions (8). The success of orthognathic surgery depends on the accuracy of the treatment plan. In the SFA, close collaboration, accurate planning and modeling are especially important due to the absence of orthodontic decompensation. Conventional surgical planning (CSP) utilizes 2D cephalometric analysis as well as photographs, dental casts, and model surgeries (21).

Knowledge gap and objective outcome

This reported case demonstrates the use of the SFA in the management of a patient with achondroplasia and seems to be the first known case report of its kind. It also demonstrates the efficient coordination of an orthodontic and oral and maxillofacial surgery team and the effective use of new technologies in treatment planning. We present this article in accordance with the CARE reporting checklist (available at https://fomm.amegroups.com/article/view/10.21037/fomm-23-9/rc).

Case presentation

A 19-year-old male with achondroplasia presented at the University of Alabama Birmingham School of Dentistry with the chief complaint that "he needed to eat and chew better". The patient said that in addition to his difficulty in incising and chewing foods, he also had speech problems related to his jaw position. Clinical examination revealed a concave facial profile, hypoplastic midfacial soft tissue, and adequate distance between the throat and chin. His malocclusion was complicated by a significant anterior crossbite with −9 mm overjet, left side posterior crossbite, and significant deep overbite. The maxillary and mandibular dental midline was coincident with the facial midline. There was no popping, clicking, or crepitation of the temporomandibular joints. No mandibular displacement of the lower jaw was noted. Extra and intra-oral photographs together with upper and lower digital impressions were taken. Furthermore, lateral cephalometric, panoramic radiographs and cone-beam computed tomographic (CBCT) radiographs were obtained (*Figure 1*).

Dental casts revealed anterior and left side posterior crossbites. On the left and right sides, the first molars and the canines presented with a Class III relationship. Cephalometric analysis revealed a retrognathic maxilla and prognathic mandible. The growth pattern was normo-divergent and skeletal growth was complete. The mandibular incisors were retroclined, the occlusal plane was slightly canted and there was moderate crowding of 5.5 mm in the upper arch. Lateral cephalometric analysis revealed severe maxillary hypoplasia with dental compensation.

All procedures performed in this study were in accordance with the Helsinki Declaration (as revised in 2013), and the study was approved as an exempt study by the University of Alabama at Birmingham. Written informed consent was obtained from the patient for publication of this manuscript and any accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

Treatment objectives and alternative

The patient was diagnosed with a significant congenital maxillary hypoplasia and mandibular prognathism. The dental, skeletal, and soft-tissue treatment objectives for this patient were to create a well-balanced facial proportion, improve dental occlusion and restore chewing function.

In the state of Alabama, patients presenting with craniofacial deformities receive state supported funding for orthognathic surgery and orthodontics up to the age of 21 years. As the patient was almost 20 years old, the team did not have a significant amount of time for treatment before the age limit was reached. As a result, the team decided on the most expeditious treatment plan to complete

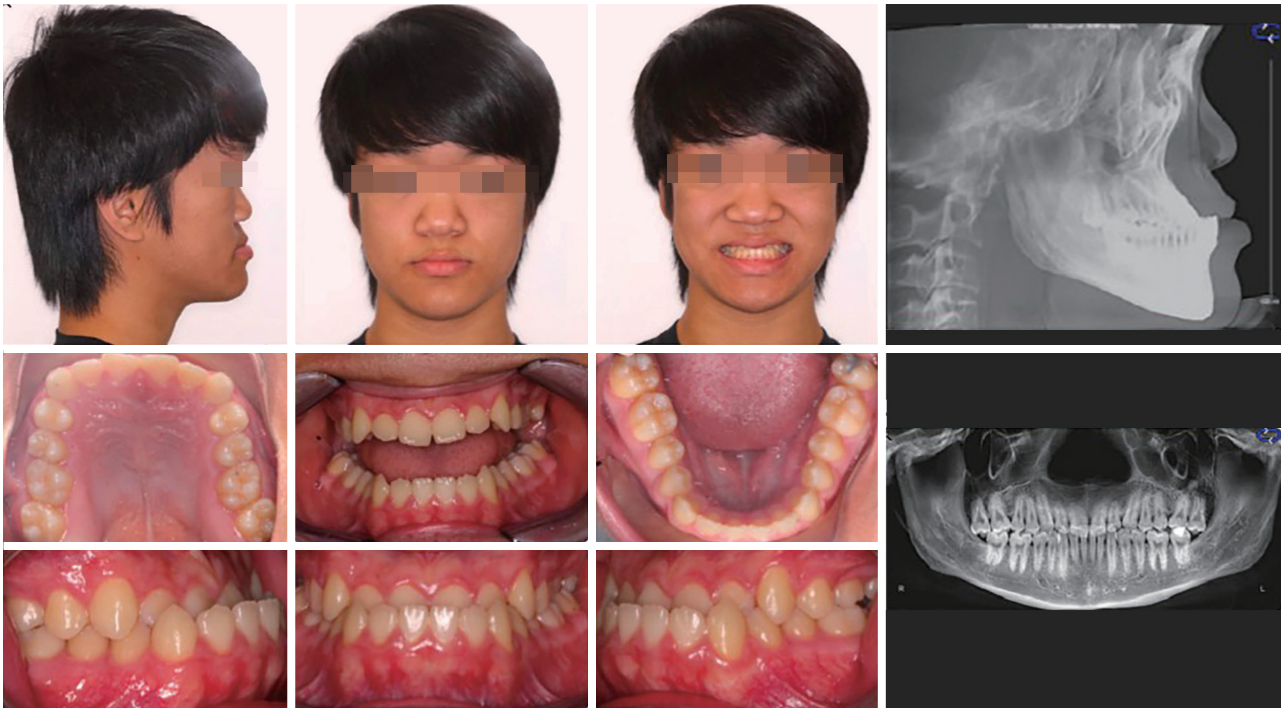


Figure 1 Composite pictures representing the patient together with radiographs representing panoramic and cephalometric views. These images are published with the patient's consent.

the treatment within the allowable funding time frame.

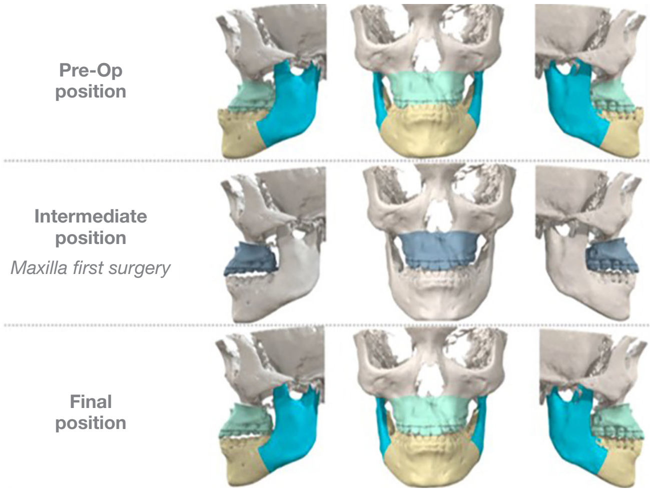
The final treatment plan was to carry out a SFA followed by orthodontic. treatment to rehabilitate the patient. The patient proceeded with Orthognathic Surgery on December 12, 2019 and all treatment was completed by May 2020. The definitive plan included non-extraction treatment, followed by a Le Fort 1 maxillary osteotomy to advance the maxilla, bilateral sagittal split setback with intermaxillary fixation screws. Post-surgical orthodontic treatment with the use of conventional orthodontic brackets was carried out to correct inter-arch discrepancies after the surgery.

The alternative plan for the patient would have been an extensive pre-orthodontic treatment to align the occlusion followed by orthognathic surgery. This treatment would include a course of post-orthodontic detailing and settling of the dental occlusion. The alternative plan was rejected because the patient wanted the state to financially support the treatment.

Treatment progress

In order to expedite treatment time, all of the surgical planning was done using the IPS Orthognathic Platform by KLS Martin (Jacksonville, FL, USA) (*Figure 2*). Both the orthodontist and oral & maxillofacial surgeon worked closely to determine the surgical movements and post-surgical orthodontic occlusion during the virtual treatment planning sessions. The treatment planning required previous orthognathic experience of the team and also the flexibility to be able to work with only what the patient presented with at the time. Once the treatment plan was finalized, the surgical splints were printed and sent to the office in preparation for the surgery.

To aid the surgeon during the orthognathic surgery, the orthodontist (C.H.K.) placed conventional metal orthodontic brackets (American Orthodontics Master Series-MBT 022 slots, Sheboygan, WI, USA) onto the maxillary and mandibular dentitions one day before surgery. An active 018 Niti orthodontic wire was tied into each arch. Orthognathic Surgery was carried out on the maxillary and mandibular jaws the day after and the patient had an occlusal placed wire to the dentition for 4 weeks. On the 4th week, the orthodontist removed the surgical splint and retied the 018 Niti orthodontic wires. In addition, the patient was instructed to use "short" anterior triangular elastic (3/16 4Oz elastics) at night times only (*Figures 3,4*). After 3 weeks,



Landmark	A-P	Right-left	Down-up
Anterior nasal spine	6.0 mm anterior	0.0 mm	0.0 mm
A-point	6.0 mm anterior	0.0 mm	0.0 mm
Upper incisor midpoint	6.0 mm anterior	0.0 mm	0.0 mm
Upper canine cusp left (U3L)	6.0 mm anterior	0.0 mm	0.0 mm
Upper canine cusp right (U3R)	6.0 mm anterior	0.0 mm	0.0 mm
Upper molar cusp left (U6L)	6.0 mm anterior	0.0 mm	0.0 mm
Upper molar cusp right (U6R)	6.0 mm anterior	0.0 mm	0.0 mm
Lower incisor midpoint	2.1 mm posterior	0.1 mm right	5.2 mm down
Lower molar cusp left (L6L)	1.8 mm posterior	0.2 mm left	2.8 mm down
Lower molar cusp right (L6R)	2.5 mm posterior	0.2 mm left	2.7 mm down
B-point	4.0 mm posterior	0.1 mm right	4.8 mm down
Pogonion	5.5 mm posterior	0.2 mm right	4.8 mm down
Menton	6.2 mm posterior	0.2 mm right	4.2 mm down
Gonion left	2.7 mm posterior	0.4 mm left	0.8 mm down
Gonion right	3.3 mm posterior	0.4 mm left	1.0 mm down

Figure 2 Surgery first planning using IPS Orthognathic software from KLS Martin (Jacksonville, FL, USA). Movements in millimeters together with 3D rendering of the skeletal changes.

Figure 3 Intra-oral splints in place after the "surgery first" orthognathic surgery. The "Initial 018" nickel titanium wire in place with the post-surgical occlusion is evident at this stage of treatment. These images are published with the patient's consent.

Figure 4 A significant change to the dental occlusion 3 months into treatment. These images are published with the patient's consent.

the orthodontist advanced the wire size to a 016×22 Niti working wire. Finally, upper and lower 18×25 TMA wires were used to detail and finish the treatment. During the course of the treatment, care was taken to maintain the anterior posterior relationship of the established surgical result and not to create too much decompensation. Finally, the patient's orthodontic brackets were debonded and orthodontic Hawley retainers were given to the patient.

At the end of treatment, a Class I molar and canine relationship, normal overbite and overjet were achieved with coincident dental centerlines to the mid-sagittal plane. In less than 6 patient visits after surgery (approximately 6 months), the treatment was completed despite clinic closure during the COVID-19 pandemic period. Final facial photographs, CBCT, and cephalometric superimpositions with initial cephalogram, were carried out (*Figures 5,6* and *Table 1*).

Discussion

While SFA has been documented for correction of severe dentofacial deformities including cleft lip and palate, severe malocclusions, and hemifacial microsomia, none have described a case with achondroplasia using our approach (22-24). This technique represents a breakthrough in the management of patients with significant facial deformities and the effective use of precious financial and clinical resources.

As mentioned previously, conventional approaches to surgical-orthodontic treatment require three main stages: preoperative orthodontics, surgery and postoperative orthodontics (8). This approach has its drawbacks including lengthy orthodontic treatment (15–24 months preoperatively; 7–12 months postoperatively). Pre-operative

Figure 5 Final clinical composites pictures and radiographs showing the panoramic and lateral cephalometric views. These images are published with the patient's consent.

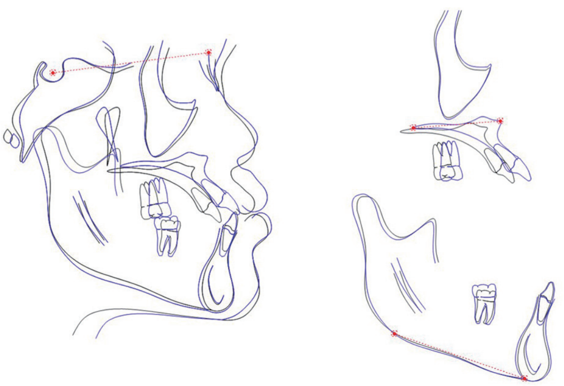


Figure 6 Initial and final cephalometric superimpositions showing the skeletal changes. The black lines indicate the pre-surgical tracing and the blue lines the postoperative and final tracings.

orthodontics exacerbate a prognathic mandibular profile in skeletal class III patients worsening the mid-treatment facial aesthetics (8). Doing so may undermine the patients' perceived quality of life violating patient-centered treatment objectives (25-28). For select patients, the SFA approach eliminates the presurgical orthodontic phase, thereby reducing treatment duration and improving satisfaction (8,29). Evidence suggests that because the SFA may provide a higher quality of life for patients because it minimizes social isolation brought on by incisor decompensation. Orthodontics is often viewed by patients as the least pleasant aspect of treatment (27). The SFA may improve patient satisfaction by reducing total treatment time (mean total duration of 14.2 months) (20). The reason for this is two-fold. First, the proper alignment of the jaws results in physiologic movement. Second, the regional acceleratory phenomenon (RAP) helps in moving teeth quickly for treatment completion (8,20). While evidence strongly supports reduced treatment times, values vary from nearly two-fold to only a 5-months (30).

The success of orthognathic surgery depends on the accuracy of the treatment plan. In the SFA, close collaboration, accurate planning and modeling are especially important due to the absence of orthodontic decompensation. CSP utilizes 2D cephalometric analysis as well as photographs, dental casts, and model surgeries (21,31). Recent advancements in technology particularly 3D printing, 3D CBCT imaging and virtual surgical planning (VSP) have improved precision and efficiency of orthognathic surgery allowing the team to utilize their

Table 1 Cephalometric measurements at pretreatment

Group/measurement	Pre-treatment	Post-treatment	Normal
Skeletal			
Sella-nasion-A point (°)	73.9	81.7	82.0
Sella-nasion-B point (°)	89.0	88.3	80.9
Point A-nasion-B point (°)	–15.1	–6.6	1.6
Skeletal vertical			
Mandibular plane—sella-nasion (°)	25.1	26.6	33.0
Frankfort horizontal plane—mandibular plane angle (°)	26.4	29.2	23.9
Dental			
Overjet (mm)	–9.0	2.0	2.5
Overbite (mm)	8.0	1.8	2.5
Upper incisors—sella nasion (°)	124.7	122.8	102.8
Upper incisors—nasion-A point A (mm)	–16.8	15.0	4.3
Lower incisors—nasion-point B (mm)	4.6	4.4	4.0
Lower incisors—mandibular plane (°)	85.4	86.0	95.0
Soft tissue			
Lower lip to E-plane (mm)	4.5	5.6	–2.0
Upper lip to E-plane (mm)	–1.6	2.7	–6.0

experience to achieve better results (32,33). Through 3D CBCT, CAD/CAM, and VSP, clinicians are now able to collaborate, preplan osteotomies, and produce cutting guides and splints all in a single web-based workflow (32). Results from various studies support time savings during pre-operative planning and the operative procedure, as well as, decreased hospitalization times (32). In addition, clinicians can also plan and communicate even when the clinical location are many miles apart (34). VSP benefits practitioners by providing a 3D view of the procedure (32). Additionally, patients benefit by increasing informed consent (32). While VSP is more expensive than CSP, a formal cost-benefit analysis has yet to be performed. 3D printing has been used to create occlusal splints, cutting and positioning guides, fixation plates/implants, spacers, and 3D printed models (35). As modern medicine continues to embrace patient-centered treatment, technological advances such as these create the potential for the SFA to become the standard in orthognathic surgery (29).

Some authors have mentioned that SFA could result in unstable postoperative occlusion and has greater potential for relapse compared with the conventional approach. As a result, care needs to be taken into account in the retention phase (20). In this case, the postoperative occlusion was less than ideal. However, through the use of orthodontic techniques, selective use of inter-maxillary elastics and vertical anchorage control, a very satisfactory outcome was achieved.

Conclusions

The SFA in patients with significant craniofacial deformity is a treatment alternative to consider especially in cases where maturity of the patient is complete and when treatment is urgent due to state financial constraints. This case report highlights the multi-disciplinary approach and careful management of such patients.

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Footnote

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Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the Helsinki Declaration (as revised in 2013). The study was approved as an exempt study by the University of Alabama at Birmingham. Written informed consent was obtained from the patient for publication of this manuscript and any accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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