

Achondroplasia: Orocraniofacial Features and Orthodontic-Surgical Management Guidelines Proposal

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Abstract: In this study, the authors aimed to describe orocraniofacial features and to suggest orthodontic-surgical managements in achondroplasia, based on a literature review. The authors focused on skeletal and dentoalveolar malocclusion in order to highlight the place of orthognathic surgery, based on our experience of 3 patients. Maxillary hypoplasia in achondroplasia typically results in an Angle class III malocclusion with an anterior open bite. The other orocraniofacial features include enlarged calvarium, prominent forehead and frontal bossing, midface hypoplasia, elongated lower face and saddle-shaped nose due to lack of development of the nasomaxillary complex.

All our patients had a typical facial appearance but each of them had their own particularities regarding medical history and severity of the dentoskeletal dysmorphism. Two of them were successfully treated by orthognathic surgery; the other declined surgical treatment and underwent orthopedic treatment only (anchorage plates). The treatment failure of this 3rd patient raises the question of the efficiency of anchorage plates in achondroplasia. In the light of the literature and our results, the authors conclude the need for personalized management based on age, medical history, severity of the dentoskeletal dysmorphism, functional and/or esthetic disorders, and the patient's needs and requests. In any patient, orthodontic management should be initiated at an early age, and orthognathic surgery modalities should be personalized and adapted to each situation.

Key Words: Achondroplasia, orocraniofacial, orthognathic surgery

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Achondroplasia is a genetic disease impairing cartilage and bone formation. It is the most common cause of dwarfism with an incidence estimated to 1/25,000.¹ In more than 95% of diagnosed patients, a mutation in the gene encoding fibroblast growth factor receptor type 3 (FGFR3), resulting in the amino acid substitution (G380R), is found. Prenatal diagnosis and preimplantation genetic

diagnosis analyses can be proposed, after appropriate genetic counseling.^{2,3}

Typical clinical manifestations are found with: short stature, shortening of the arms and legs, short stubby trident hands. The most common orocraniofacial characteristics are relative macrocephaly, depressed nasal bridge, maxillary hypoplasia, and malocclusion.⁴

The American Academy of Pediatrics Committee on Genetics outlined recommendations for management of children with achondroplasia.⁵ Patients require specific management regarding maxillofacial issues in order to allow occlusal restauration and to avoid breathing and speech disorders, but also, to offer functional and esthetic benefits.

However, there is poor literature on orthognathic surgery in achondroplasia.

The aim of our article is to describe orocraniofacial features and suggest general managements in achondroplasia, based on a review of the literature, and to focus on skeletal and dentoalveolar malocclusion in order to highlight the place of orthognathic surgery, based on our experience of 3 patients.

MATERIALS AND METHODS

Systematic Review

We searched across all studies specific to orthodontic and maxillofacial management of achondroplasia performed with the help of PubMed interface of Medline by searching the keywords: “Achondroplasia” conjugated to “orthodontic” and “maxillofacial surgery.” We included the most relevant ones and we pushed the investigations to the bibliographic references of the included articles. We excluded all studies that had not been written or accurately translated in either English or French language.

Our Experience: 3 Clinical Reports

Our study was based on all achondroplasia patients managed in the Maxillofacial Surgery and Stomatology Department at Lille University Hospital between January 1997 and January 2018.

This series of 3 patients was thus found with the cooperation of the hospital's Medical Information Department.

The study was approved by the local ethics committee at the University of Lille.

RESULTS

Main Features of Achondroplasia General Clinical Signs

Every specialized practitioner should be aware of the main features of achondroplasia, to avoid misdiagnosing the condition (Table 1).

The general characteristics of achondroplasia include:

- a disproportionate short stature. Medium adult heights are 131 ± 5.6 cm for males and 124 ± 5.9 cm for females⁶;

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TABLE 1. Major Clinical and Radiological Features of Achondroplasia

General	Disproportionate short stature Rhizomelic shortening of the extremities Shortening and thickening of the long bones Short, broad, and cupped phalanges Short stubby hand with trident configuration
Craniofacial	Enlarged calvarium Large frontal sinuses Prominent forehead Midface hypoplasia Nose saddle shaped Concave facial profile
Oral	Anterior open bite Class III or Class I malocclusion

- rhizomelic shortening of the extremities in contrast to an average-sized trunk;
- short stubby hand with trident configuration (increased space between the third and the fourth fingers);
- prominent buttocks and a protuberant abdomen;
- lumbar spine stenosis and lumbar lordosis (curved lower spine);
- thoracolumbar kyphosis is frequent, related to hypotonia, flat chest, and protuberant abdomen;
- joints laxity, especially in knees, and limbs deformation, such as limited elbow extension, inferior dislocation of the shoulder, bowlegs deformity. Lower extremity deformities include coxa vara, genu varum, and heel varus⁴; and
- normal intelligence with independent and productive lives unless hydrocephalus or other central nervous system complications occur.²

The most commonly reported complications in patients are:

- cervicomedullary compression often related to a stenosis of the foramen magnum⁷;
- otolaryngeal system dysfunction with recurrent otitis media, adenotonsillar hypertrophy⁸; and
- obstructive sleep apnea.⁹

General Radiographic Signs

The radiological features include:

- shortening and thickening of the long bones with metaphyseal flaring and cupping;
- phalanges are short, broad, and cupped;
- iliac bones are short and rectangular with narrow sacroiliac notches and short, wide pubic and ischial bones;
- bullet-shaped vertebral bodies with posterior scalloping and narrowing of lumbar interpedicular distances; and
- bullet-shaped vertebral bodies with posterior scalloping and narrowing of lumbar interpedicular distances.²

Orocraniofacial Clinical Signs

The orocraniofacial features include:

- enlarged calvarium with relative macrocephaly, can lead to hydrocephalus;
- prominent forehead and frontal bossing;
- midface hypoplasia, collapsed midface and a deficient maxilla, and an elongated lower face;
- depressed nasal bridge, saddle-shaped nose because of lack of development of the nasomaxillary complex¹⁰; and
- concave profile.

In particular, occlusal features include:

- anterior open bite;
- Class III malocclusion¹⁰⁻¹²;
- Class I malocclusion¹³⁻¹⁵;
- Class II division 1 malocclusion with protrusive maxillary incisors¹⁶;
- mouth breathing¹⁷; and
- macroglossia.¹⁰

Cephalometric Signs

- enlarged calvaria;
- frontal bossing;
- large frontal sinuses;
- occipital prominence;
- normal anterior cranial base length, strikingly shortened posterior cranial base length, an acute cranial base angle;
- short upper facial height, recessed maxilla, posterior tilt of the nasal floor, and a prognathic mandible that is anteriorly displaced but of normal size with a normal gonial angle commonly contributing to skeletal class III pattern of malocclusion
- short nasal bone that was deformed and depressed;
- short upper facial height, recessed maxilla;
- posterior tilt of the nasal floor; and
- high coronoid process.¹⁸

Clinical Reports

Our study is based on 3 patients suffering from achondroplasia. Maxillofacial and orthodontic management took place at the Maxillofacial Surgery and Stomatology Department of Lille University Hospital.

Diagnosis was made previously, suspected during obstetrical follow-up and confirmed at the time of birth and patients were sent to us, for specialized management, after puberty.

There was a female aged 21 and 2 males aged 19 and 18 at the time of our study, included between December 2009 and January 2018.

Patient 1

Female subject, she consulted at the age of 12. Typical manifestations of achondroplasia were observed. Extraoral examination revealed severe midface hypoplasia, concave facial soft-tissue profile, and vertical chin excess. Intraoral assessment showed Class III molar relationship (Figs. 1-4).

Orthodontics procedures led mainly to the preparation for orthognathic surgery with transverse maxillary expansion, alignment, leveling, and lifting of compensations. Surgical management began at the age of 15 with the placement of a palatal circuit breaker. This allowed a surgical cross-sectional expansion distraction in order to avoid any tooth extraction. After puberty, at the age of 17 years, bimaxillary orthognathic surgery was performed with Le Fort I advancement osteotomy, bilateral sagittal split osteotomy, and reduction genioplasty. The procedure provided a satisfactory occlusion in Class I. The profile was improved. The final orthodontic precisions improved occlusion on the left side where there was a slight infarction on the canine and premolar area.

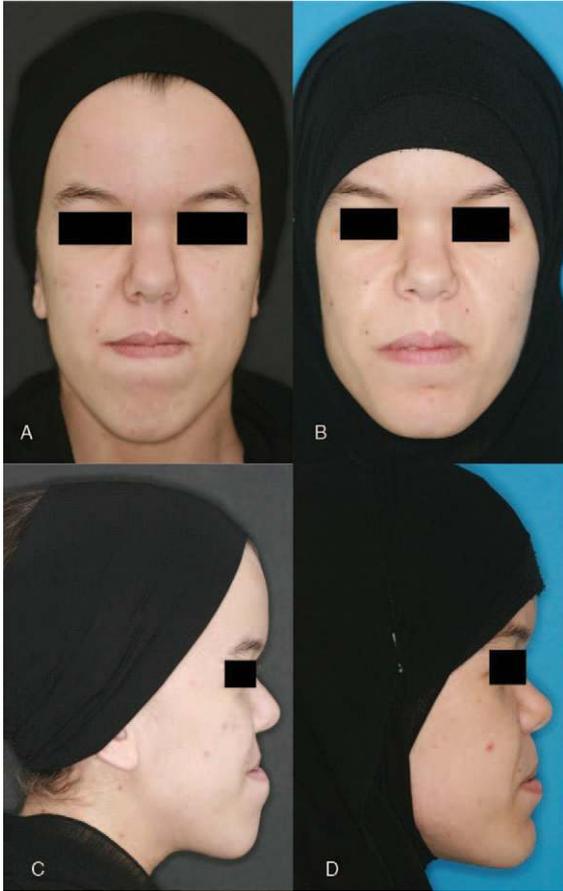


FIGURE 1. Patient 1. (A) Presurgical frontal view showing typical facial appearance. (B) Postsurgical frontal view (note vertical chin excess reduction). (C) Presurgical profile view showing typical concave facial profile. (D) Postsurgical profile view showing profile improvement.

Patient 2

Male subject, he consulted at the age of 16. Medical history revealed obstructive sleep apnea by upper airway obstruction managed by midface advancement using Le Fort III distraction osteogenesis technique, at the age of 6. General physical examination showed the classic features of achondroplasia. More precise examination revealed concave profile, Class III molar relationship with anterior open bite. Dentoskeletal dysmorphism was less pronounced given the history of midface advancement in childhood. He had taken up orthodontic treatment by alignment and leveling, in his hometown, since the age of 12. His main concern was the inability to bite with his anterior teeth.

Surgical management began at age 17, after puberty, with bimaxillary orthognathic surgery with Le Fort I advancement osteotomy and bilateral sagittal split osteotomy. A very acceptable Class I occlusion was achieved. Upper and lower teeth were in good alignment, open bite closure was corrected. The profile was improved. An endoclasia on the right hemimaxillary was caught up by orthodontic expansion therapy.

Patient 3

Male subject, he consulted at the age of 11. On physical examination, the classic features of achondroplasia were observed. The patient manifested skeletal and dental Class III with edge to edge incisor relationship.

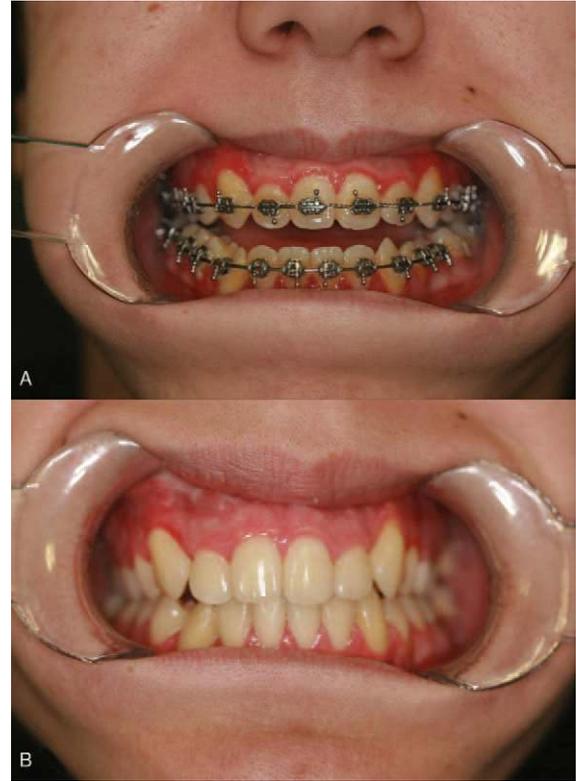


FIGURE 2. Patient 1. (A) Presurgical view of occlusion showing Class III malocclusion. (B) Postsurgical view of occlusion.

At first, we proposed an orthopedic treatment by anchorage plates. Orthognathic surgery was recommended after puberty. Therefore, the establishment of 4 anchorage plates was performed at the age of 11 years to allow continuous maxillary traction with light forces. Maxillary deficiency was treated by orthodontic expansion therapy to help the upper canines to reach the dental arch. At the age of 13 there was still a Class III malocclusion with edge-to-edge incisor relationship. Orthopedic treatment failed. However, the patient and his family decided to cancel planned orthognathic surgery together with the whole orthodontic-surgical management. Therefore, the anchorage plates were removed.

DISCUSSION

Diagnosis

Achondroplasia should be diagnosed, at the earliest in utero, at the latest at birth according to physical aspect.³

It is crucial to be aware of the main orocraniofacial characteristics in order to adequately plan the treatment and restore occlusal functional and esthetic relationships.

In addition to the above-mentioned general signs, all our patients had a typical facial appearance (Results, Table 1). Maxillary hypoplasia typically results in an Angle Class III malocclusion with an anterior open bite. This is in agreement with the literature. Indeed, the main occlusal problem in patients with achondroplasia is Class III malocclusion due to innate shortening of the base of the skull. The generalized defect in endochondral osteogenesis is due to a deficiency in cartilage cell proliferation. In long bones, the epiphyses are rapidly fused with the diaphysis that explains the short stature. This shortness of the cranial base is explained by



FIGURE 3. Patient 1. (A) Presurgical angled view of occlusion. (B) Postsurgical angled view of occlusion showing very acceptable Class I occlusion achieved—slight infarction on the canine and premolar area.

the pathophysiology of the disease that induces a premature ossification of the synchondrosis of the chondrocranium. Therefore, the viscerocranium that is just under it remains in a backward position. Despite compensations of the viscerocranium class III malocclusions are frequent. This pathophysiology explains the underdevelopment aspect of the middle third of the face. This abnormal configuration results in retraction and reduced vertical height of the maxillary too. Because condylar cartilage is the product of periosteal chondrogenesis, mandibular growth is not affected.¹⁹ The saddle-shaped nose is due to underdevelopment of the nasomaxillary complex.

Patients with Class I malocclusion have been reported.^{13–15} Ohba et al report a Class II division 1 malocclusion with protrusive maxillary incisors in a 10-year-old achondroplasia patient who demonstrated a tongue-thrust swallowing pattern.¹⁶

Skeletally, however, this patient presented with a class III pattern with maxillary underdevelopment due to achondroplasia. The only exception found is a patient with achondroplasia also presenting with oligodontia. In this patient, it is difficult to know what occlusion would have been present if teeth would have not been absent. There was no available data regarding the incidence of oligodontia in achondroplastic children. This patient had midfacial hypoplasia with retruded maxilla; however, marked mandibular prognathism was not observed.²⁰ In this patient, the finding of oligodontia was more pronounced than any other patients reported in literature. Nevertheless these observations pointed out the huge adaptation potential of the dentoalveolar processes even in patients where there is important Class III relationship of the skeletal bases.

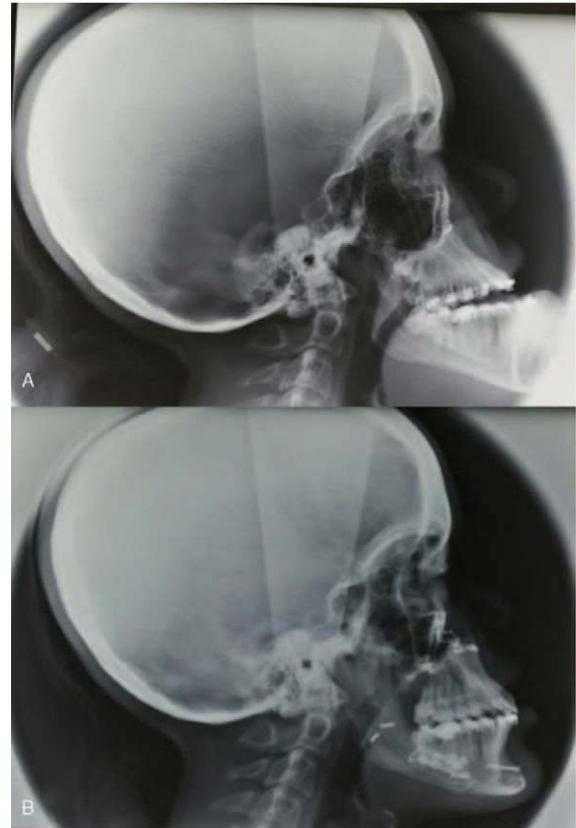


FIGURE 4. Patient 1. Presurgical (A) and postsurgical (B) profile teleradiographies showing considerable improvement with Le Fort I advancement osteotomy and bilateral sagittal split osteotomy by maxillary surgery and reduction genioplasty.

Timing of Orthodontic-Surgical Management

Concretely, the objective of orthodontic and/or surgical treatment of achondroplasia is that of a Class III by maxillary deficiency.^{12,14} It is first of all to promote maxillary growth and restrict mandibular growth. It is necessary to improve the skeletal Class III jaw–base relationship, correct the anterior crossbite and open bite, establish proper overjet and overbite, and achieve an acceptable occlusion with a functional Class I occlusion. Treatment planning for patients with skeletal deformities is often considered challenging. In the treatment of any malocclusion, the orthodontist/maxillofacial surgeon must consider many factors including the growth potential of the patient in relation to prepubertal and pubertal growth phases. In patients in which growth may be interrupted or deficient because of genetic factors, special attention must be given. Treatment modalities may be limited since “normal” growth cannot be used as a positive treatment factor. Growth potential is different precisely in patients suffering from achondroplasia; therefore, management must be specific.^{11,13} In fact, it is a question of setting up the treatment plan in an early and adapted way.

The American Academy of Pediatrics recommends a review of orthodontic problems in achondroplasia after 5 years of age.²¹ Early orthodontic evaluation should be carried out in achondroplastic patients to attempt interceptive orthodontics. Orthodontic treatment only started at 12 years old for patients 1 and 2.

Anchorage plates offer the possibility to apply pure bone-borne forces between the maxilla and the mandible for 24h/d and maximize orthopedic effects by decreasing concavity of Class III

dysmorphism profile.²² In patient 3, orthopedic treatment was done but the outcome was poor raising question on the efficiency of this treatment in achondroplasia. Indeed, these tractions are supposed to have action on the sutures of the maxilla that should not be affected by the disease so the explanation for the failure is unclear.

Orthodontic treatment should be rapidly initiated by using a palatal expansion appliance to correct the cross-bite malocclusion and to gain space. Myofunctional therapy to stop the tongue-thrusting habits should last throughout orthodontic treatment.¹² When a discrepancy between the maxilla and mandible is not severe or the skeletal deformity is not the main concern, the treatment decision to correct the malocclusion in patients with achondroplasia may be limited to orthodontic treatment only.¹¹ Early treatment is not always possible. Al Saleem et al studied an 11-year-old patient with severe maxillary hypoplasia and skeletal Class III. Orthodontic consultation was conducted revealing that it was too late to orthodontically intercept at this age, as the patient approached puberty. So orthognathic surgery was planned after complete puberty.¹⁰

Of course, despite adequate orthodontic treatment, depending on the severity of the dentoskeletal dysmorphism, it is not always possible to obtain a satisfactory result. Our series are part of this framework.

Orthognathic Surgery

Literature depicting orthognathic surgery and achondroplasia is poor. Surgical procedures usually depend on the severity of facial skeletal deformities. Analysis of the problem clearly indicates the need for moving multiple components of the facial skeleton into new positions in order to provide occlusion correction. At first sight both upper and lower midface require simultaneous movements in exact opposite directions to normalize the typical skeletal deformity. However, considering the pathophysiology it is essentially the retrusion of the midface that is the main anomaly to correct.

For severe patients with comprehensive correction using various craniofacial surgery techniques, for example, frontofacial advancement and Le Fort I and vertical subnasal osteotomy, satisfactory craniofacial function, and esthetics have been obtained. A simple combination and application of standard techniques of craniofacial surgery in achondroplastic patients allowed a very good result.²³ However, whatever the technique it must be taken into account that the main anomaly is related to the cranial-base restriction.

Possible additional surgery to address the remaining skeletal deformities should be performed after completion of active growth. The dentoalveolar component of a skeletal deformity can be handled independently of craniofacial management.²⁴ Achondroplastic patients can be treated in stages. Karpagam et al report a 14-year-old female patient with achondroplasia who presented a combination of anterior openbite, vertical maxillary excess, severe maxillary retrusion, and Class I molar relation with lip incompetence. Firstly, they suggested, correction of the dental component of anterior open bite followed by the treatment of the upper midface including the nasal complex secondly.¹⁴

In our experience, patient 2 had previously benefited from a midface advancement, during childhood, by means of a Le Fort III distraction osteogenesis for obstructive sleep apnea by upper airway obstruction.

Le Fort II and III osteotomies and distraction are performed between 6 and 10 years of age, when the craniofacial skeleton is easier to mobilize. Indications for a Le Fort III distraction at this age are obstructive sleep apnea patients with high risk for upper airway obstruction in those syndromes that include midface hypoplasia involving the nasal and zygomatic complex and bony orbits, for example, the Crouzon, Apert, and Pfeiffer syndromes.²⁵

Authors do not consider this step as part of the specific planning treatment for achondroplasia. When we treated patient 2,

obstructive sleep apnea had been resolved and Class III dentoskeletal dysmorphism was less marked than in patient 1. However, occlusal disorder and difficulty in biting with anterior teeth were 2 problems that still remained.

First, a Le Fort III distraction should be performed during childhood, and second, an additional Le Fort I osteotomy may be necessary, when skeletal maturity is reached, in order to achieve an intermaxillary relationship enabling a stable occlusion.²⁶

In both patients, the presurgical orthodontic stage resulted in satisfactory decompensation of the mandibular incisor inclination and alignment of the teeth in both arches. Teeth extractions were discussed but not planned given the stimulating effect of teeth evolution on maxillary growth. Lateral maxillary expansion produced enough space for teeth alignment, even though the patient had been treated without premolar extractions in patient 2. Maxillary narrowness and bilateral crossbite were treated with maxillary surgical expansion by distraction in patient 1. Then in both patients, by the careful combination of standards techniques, that is, Le Fort I and bilateral sagittal split osteotomy, a good correction had been obtained. Reduction genioplasty was necessary in patient 1 given the chin vertical excess. Rhinoplasty was systematically proposed but none of our patients had wanted to correct the nasal malposition. In both patients, the final occlusion was Class I with normal overbite, overjet and dental arch alignment. Fixed orthodontic appliance was placed after surgery to ensure stable results.

Midface advancement using subcranial Le Fort III osteotomy after puberty has been reported.

According to Barone, Le Fort III allows corrections in the upperface and can resolve obstructive sleep apnea.²⁷ However, in achondroplasia, it is more logic to perform a Le Fort II osteotomy. Indeed, the zygomatic bones should not be affected by the disease. Of course the planed osteotomies can vary from patient to patient. Richard showed interest in advancement orthognathic surgery for patients with achondroplasia with obstructive sleep apnea.²⁸ They were treated with either an isolated Le Fort III distraction or Le Fort II distraction with or without subsequent Le Fort I and bilateral sagittal split osteotomies. It should not be overlooked that other interventions manage to improve sleep apnea in achondroplasia, depending on the etiology of the obstruction. Sato et al showed that adenoidectomy and tonsillectomy allowed dilatation of the pharynx and improved craniofacial and pharyngeal morphologies, thereby decreasing sleep apnea.⁹

Admittedly, our approach is different since it focuses more particularly on occlusal relationships in patients who do not suffer from obstructive sleep apnea.

However, our study confirms the need for orthodontic-surgical management adapted to each patient with achondroplasia. We conclude the need for personalized management based on age, medical history, severity of the dentoskeletal dysmorphism, functional and/or esthetic disorders, and the patient's needs and requests, overall (Fig. 5).

Perspectives

In our patients, the nose was still in a retruded position. Yet, instead of a typical Le Fort I osteotomy, an intermediate Le Fort II osteotomy carrying the nasal pyramid during bimaxillary orthognathic surgery, is feasible. It involves adjusting both the occlusion and the nasal morphology but allows to avoid an extra surgical procedure.

There are advantages and disadvantages in combining rhinoplasty with orthognathic surgery. For example, maxillary advancement may widen the nasal base or shorten the upper lip. If nasal change is indicated or desired, a single surgery may be planned. However, it may be technically difficult to correct a subtle nasal malformation during maxillary osteotomy for reasons such as

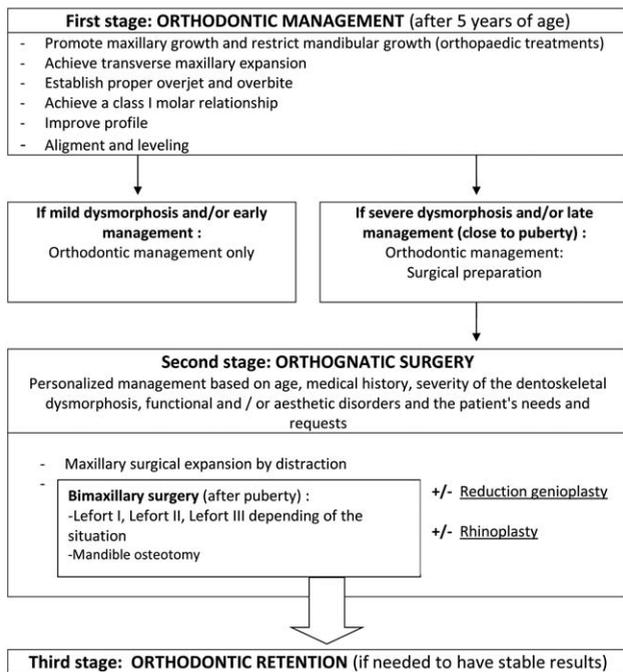


FIGURE 5. Management guidelines proposal.

intraoperative edema and performing rhinoplasty on an “unstable foundation.”²⁹ The main difficulty is to correct the nose retrusion that should be done in our conception independently of the other procedures. Indeed, when classic osteotomies are performed they do not change the discrepancy of the nose with the other part of the craniomaxilla area. If many procedures are available to solve this anomaly it is not clear which one to choose.

Meanwhile, none of our patients wished to correct nasal malposition, the main complaint remaining malocclusion. Therefore, rhinoplasty was not performed in our patients, but should be included as an option in the management plan.

The multiplicity of surgical procedures to be performed for a more satisfactory correction of the profile as well as potential perioperative complications also have to be taken into account. Dhiman et al showed that individuals with short stature skeletal dysplasias report lower health-related quality of life.³⁰ A patient with achondroplasia not only requires specific medical management, but also requires special attention with personalized management along with psychological support during surgical procedure in particular.

CONCLUSION

Achondroplastic patients may have a better quality of life when dental occlusion function and facial appearance are improved with appropriate orthodontic management and orthognathic surgical correction.

Every maxillofacial surgeon should be aware of the clinical characteristics of achondroplasia in order to adapt and to personalize patients’ management. Orthognathic surgery strategies should be set according to the patients’ age, medical history, severity of dentoskeletal dysmorphism, functional disorders, esthetic issues, and the patient’s request.

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