

# Rare diseases and orthognathic surgery



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## ABSTRACT

*Orthognathic surgery, is often required for patients with rare diseases in order to normalize occlusion that is generally severely affected. There is no one specific surgical approach because anomalies encountered in this population often require various complex technical procedures. Unlike common dental malocclusions, agenesis, skeletal dystrophies and functional problems are gathered. Orthodontic preparation, before surgery, is made difficult, resulting in a challenging treatment process. In certain cases, the stability of the result can be uncertain due to major dysfunctions. However, given the functional as well as morphological improvement that these procedures bring about, they should be planned if the dysmorphia is significant.*

## KEY WORDS

*Orthognathic surgery*

*Rare diseases*

*Dental dysplasias*

*Relapse*

## 1 – INTRODUCTION

In the European Union, a disease is considered rare when it affects less than 5 out of 10,000 people. The number of patients can however be high, since some 8000 rare diseases have been described. The majority of them are associated with genetic defects.

Rare diseases by the numbers:

- 5 new pathologies are described each week worldwide, affecting 5 to 8% of

the world population, to be specific 4 millions French people, 25 million Europeans and 27 million North Americans.

- 65% of these rare diseases are serious and disabling. They are characterized by:
  - 1) early appearance before the age of 2 in two thirds of the cases;
  - 2) chronic pain in 1 out of 5 patients;

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- 3) a motor, sensory or intellectual deficit, involving a decrease of autonomy in 1 out of 3 cases;
- 4) a fatal prognosis in half the cases, which is why 35% die before the age of 1, 10% between the ages of 1 and 5, 12% between 5 and 15 years old.

The terms "rare disease and orthognathic surgery" imply the existence of a "disease" (a recognized pathologic entity), that is rare in nature and associated with a malocclusion directly related or not to this disorder.

In fact, it is difficult to determine exactly how many pathologies fall in this category. In reality, the term "rare diseases" includes a considerable number of different physiopathologic situations even though the number of cases for each one of them is limited.

Although the anomalies listed as the cause for a range of clinical situations are complex, many of these malformations are certainly the result of initial malformative anomalies that cause secondary deformations leading to dysfunctions<sup>1</sup>. These "malfor-

mation/deformation" interactions are the reason we encounter clinical situations where it becomes difficult to determine whether problems stem from the malformation or from complications related to the deformation. In spite of all our physiopathological understanding, it is important to know what the *primum movens* is, in order to suggest a treatment that doesn't simply rely on some physiopathological data<sup>1</sup>.

Given the diversity of rare diseases, presenting an article on orthognathic surgery as a treatment becomes an even greater challenge since the various clinical cases involved are also quite diversified. An examination of the bibliography was also rather straightforward, since we did not find a single article dealing with this subject in general manner. However, the bibliography contains articles that describe orthognathic surgery for various syndromes when they as such present specific entities.

In this article, we will therefore lay out the general rules of orthognathic treatment plans for rare diseases.

## 2 – SPECIFICITIES OF MALOCCLUSIONS ENCOUNTERED WITH RARE DISORDERS IN PEDIATRIC DENTISTRY

A rare disease can have a major impact on the orofacial complex. Rare diseases lead to a number of problems that appear during embryonic development, and manifest in various ways. From a strictly dental point of view, the anomalies encountered involve number, shape and structure of the teeth. Certain pro-

blems can have an impact on both dentitions or on only one. Some endocrine problems, and problems involving phosphate metabolism or vitamin deficiencies become additional concerns during postnatal development. Locally, malocclusions can also occur because of mechanical accidents, either premature or

delayed developmental eruption or functional problems: low muscle tone problems, or incorrect lingual posture and/or major functional problems in the orofacial complex (breathing, swallowing, mastication and phonation).

Patients with facial asymmetry and/or gothic palate are prone to crossbite: this malocclusion, that is also found in the general population, can be bilateral or unilateral, and in most cases is associated with a deviation of the incisor midlines. Correcting in the deciduous dentition will foster neuromuscular development and functional equilibrium whereas withholding treatment incurs the risk of establishing asymmetric muscular activity and exacerbating the insufficient development of the hemiface.

Children with rare diseases often present sequelae including disrupted dental development (either premature or delayed) and ectopic eruption that may or may not cause temporary malocclusion. These symptoms are frequently accompanied by microdontia or macrodontia that further complicate occlusal problems.

The number, shape and position of teeth are even more problematic anomalies and they contribute to the appearance and complications of malocclusions: when these anomalies are detected late, especially if the deciduous dentition is correct, they "quietly" further the establishment of malocclusions that drastically disrupt neuromuscular interplay, impede maturation of the functions, progressively modify the appearance of the face and significantly impact the psychological equilibrium of the child and of the adolescent.

With rare diseases, there is great intra and interfamilial variability in anomalies of number of teeth.

Hyperodontia involves the persistence of deciduous teeth in the arch into adolescence or adulthood and permanent teeth remain in position or are impacted whether or not they are misshapen.

Hyperodontia may affect the mandible or the maxilla, temporary or permanent dentition or both. Subsequent malocclusions are analogous to those encountered with oligodontia that are the most common and are associated with ectodermic dysplasias with more than 170 clinical variations.

Anodontia may involve only one of the two dentitions or only one of the two maxillaries with conical incisors on the other. Certain ectodermic dysplasias include anodontia of permanent teeth although all the temporary teeth are present. When the deciduous teeth are present, they are small and conical with gaps in between them (Fig. 1).

In addition to congenitally missing teeth and anomalies in shape, some rare diseases are associated with



*Figure 1*  
*Appearance of conical teeth.*

anomalies in structure. Both dentitions may be affected to varying degrees: there are sometimes defects in amelogenesis (Fig. 2) and extent of damage varies, and can be different for two neighboring teeth even for children from the same family. There may also be *dentinogenesis imperfecta*, that associated with *osteogenesis imperfecta*, often includes a Angle's Class III malocclusion, a posterior cross-bite and an anterior open bite. It should be noted that the more extensive the *osteogenesis imperfecta*, the greater the severity of the malocclusion.

"Ghost teeth" are a characteristic of odontoblastic dysplasia where the mesodermic and ectodermic tissues are damaged. The asymmetric expression of the pathology that generally affects a hemiarch, where there are often dysmorphic buds (Fig. 3), a persistence of primary teeth whose progressive wear fosters the eruption of the teeth from the hemiarch of the same side, and would exacerbate the malocclusion.



Figure 2  
Amelogenesis  
*imperfecta*.



Figure 3  
Dystrophic buds.

### Certain anomalies are found in all these diseases

Regardless of the cause of these malocclusions with anomalies in shape, structure or number, the consequences can be the same: a decrease in number or an alteration of the dental organs will create problems for maintaining the vertical dimension of occlusion.

Missing or defective deciduous teeth, insufficient substance or structural weakness of the dental tissues cause losses ranging from the abrading of the surface of the teeth and even the almost total disappearance of the anatomical crown; periapical lesions are often the consequence of necrosis and are the source of early avulsion of the teeth with subsequent loss of bone or arch length that further complicate the malocclusion factors and contribute to their aggravation.

Rapid abrasion or attrition will create defects in the occlusal relationships that lead to additional, premature or later wear and new levels of the occlusal planes depending on the nature of the dental expression of the disease.

Ectopic eruptions or blocked eruption of permanent teeth into the arch as well as the abnormal persistence of temporary teeth that have very thin enamel with little resistance to the prolonged masticatory forces will entail wear with loss of vertical dimension.

Rare diseases with dental anomalies have in common loss of vertical dimension and a lack of posterior support: The child will tend to protrude the mandible forward and will develop a functional class II or on the

contrary will exacerbate a preexisting Class II.

The consequences will be functional, esthetic and psychological: the child will have, along with masticatory problems, the profile of a "small old person", with an everted lower lip and an alteration of the lower part of the face, particularly visible during growth spurts; and the adolescent, who already feels awkward with this pathology, will tend to worsen the

effect of disequilibrium by becoming withdrawn, by lowering his head and scrunching his neck. The treatment plan will depend on the severity and nature of the lesions as well as how the patient is handling the situation: the treatment should provide a solution for the disruption of the occlusion and for the social embarrassment that comes from disturbances affecting mastication, speaking or esthetics.

### 3 – FEATURES OF ORTHODONTIC TREATMENT PREPARATION FOR RARE DISEASES

Maxillo-facial surgeons send patients suffering from maxillo-facial syndrome to consult with an orthodontist so that they may receive essential orthodontic preparation that precedes orthognathic surgery.

An orthodontic treatment plan is problematic due to a number of factors:

- the existence in certain cases of cleft lip and palate with associated dental anomalies;
- the presence of dental agenesis, supernumerary teeth or specific pathologies of the periodontal ligament making dental movements very difficult;
- the importance of particularly significant dental compensations in some deforming syndromes;
- the legitimate request for esthetic treatment from patients and their parents;
- the management of cleft lip and palate.

A cleft lip and palate is managed surgically starting from the first months of the infant.

But this cleft has a number of implications: dental agenesis, dental misalignment, maxillary transverse deficiency, management of small and large fragments, impacted teeth, limited bone volume that complicates the eruption of impacted teeth (especially the canines).

The side effects of cleft lip and palate should be managed by conventional orthodontic procedures for maxillary transverse expansion (screw expander, quadhelix), dental alignment with a multi-bracket appliance, eruption of impacted teeth by orthodontic traction.

It is difficult to bond orthodontic appliances since brackets and bands must be adapted to dental substance that is often insufficient.

#### Dental agenesis

Dental agenesis is frequently encountered in deformative syndromes. These agenesises will complicate the important task of dental alignment because the practitioner

must manage the space left by the missing tooth for a future prosthetic rehabilitation, they will complicate the treatment plan because the orthodontist must decide whether or not to remove the contralateral teeth (depending on the occlusal and skeletal relationships), they will complicate the bonding of appliances given the insufficient volume of dental substance. Finally, in certain cases, there are some ligament disorders that prevent "normal" movement of the teeth.

### Dental reductions

As in all cases of significant bone deficiency, whether saggital, transverse or vertical, there are compensatory dento-alveolar movements.

For patients who are treated early, these compensatory movements are more or less pronounced. They will have to be corrected in order to recover dental inclinations that are compatible orthognathic surgery (arch coordination).

### Esthetic requests

Obviously, the esthetic consequences of deformative syndromes

are significant. Requests for esthetic "rehabilitation" is of the utmost importance for parents and for patients as they mature, continue their education and are seen by others.

From an orthodontic standpoint, it would be advisable to use discreet appliances (palatal plates, ceramic brackets), to mask the spaces created by missing teeth with pontics and to achieve dental alignment with faster treatment in order to make the smile more esthetically pleasing.

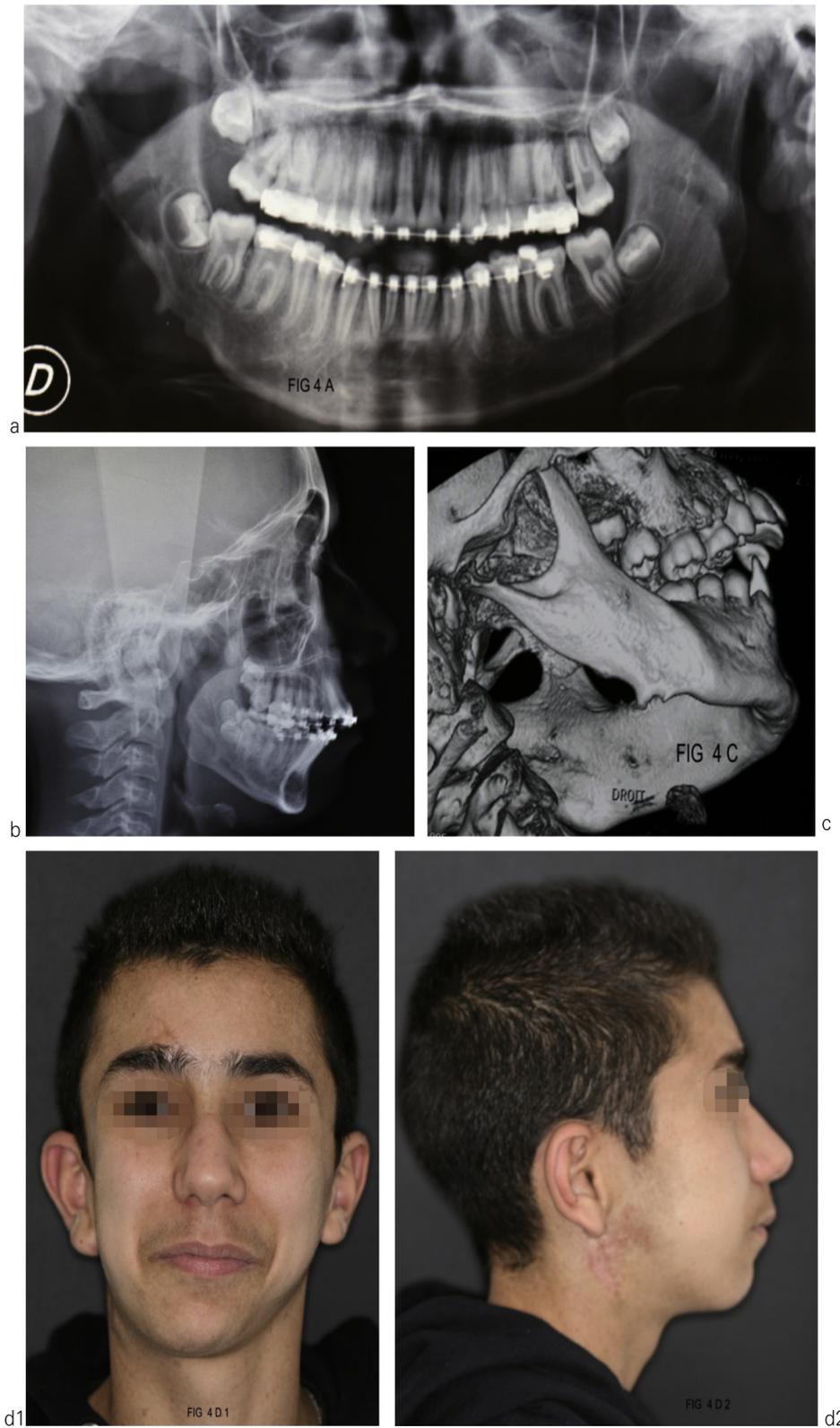
These patients do not represent the majority of cases encountered in "big cities". However, every orthodontist should be aware of technical and mechanical procedures to use in these cases, including the many challenges that are an integral part of this type of treatment plan.

The psychological factor is quite significant for these patients who early on are leading an "over-medicalized" existence. The practitioner has to show patience and support in order to have the patient accept a long and complicated treatment. These orthodontic procedures are performed in the context of a multi-disciplinary therapeutic plan where all the specialists work with one another and with the patient.

## 4 – ORTHOGNATHIC SURGERY

In this case more than in others, orthognathic surgery must take into account functional problems in order to restore occlusion for patients who have occlusal loss because of some disorder.

Orthognathic surgery is not any one specific technique but actually has to be adapted to the anomaly that must be corrected. Surgeons obviously use some traditional techniques and are fully aware that since



*Figure 4*  
 Young patient presenting Von Recklinghausen disease with significant resorption of the ascending ramus, of the angle and of the right condyle. Orthognathic surgical procedure including a maxillo-mandibular osteotomy with genioplasty and cranial bone graft to consolidate the ascending ramus.  
 a: Preoperative panoramic xray.  
 b: Lateral cephalometric xray.  
 c: Mandibular computed tomography.  
 d 1 and 2: Facial and lateral views of the patient.



Figure 4

*e 1 and 2: Postoperative view of the patient.*

*f 1 and 2: Postoperative occlusion.*

*We observed deterioration of the result in part related to the skeletal realignment of the ascending ramus of the mandible in spite of the bone graft surgery.*

the dysmorphisms are often significant, they then must necessarily resort to a particular technique or to bone grafts (Figs. 4, 5). In spite of the challenges that should not be minimized, there is a wide range of techniques in the arsenal of orthognathic surgery so that it is almost al-

ways possible to choose a procedure that will easily correct the observed deformity.

Sometimes, orthognathic surgery cannot be performed due to some major risk, from anesthesia or from the possibility of worsening the disorder (Fig. 6).

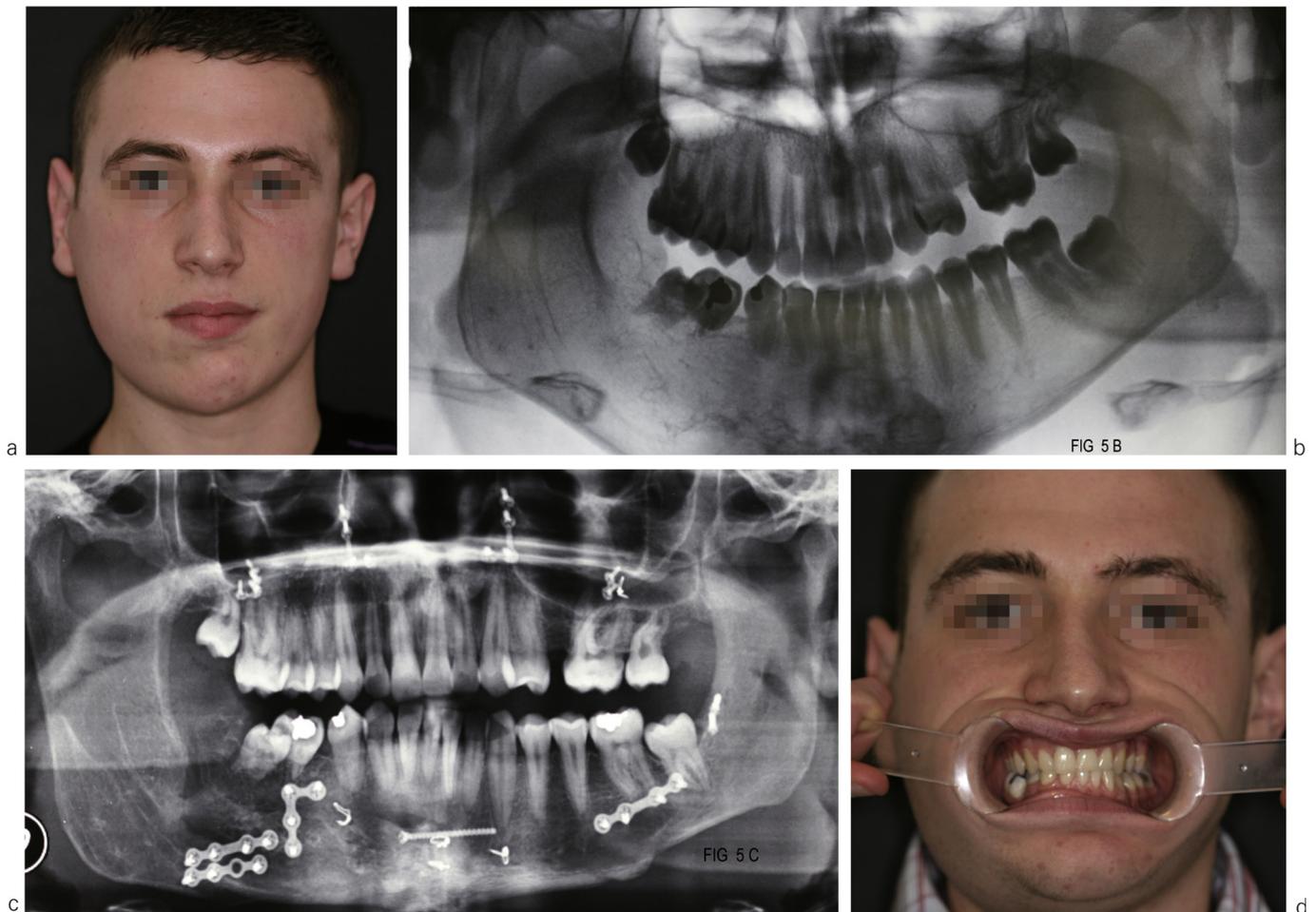


Figure 5

Right facial hemi hypertrophy with osseous and dental dysplasia located on the right.

a: Facial view, notice the right facial hypertrophy.

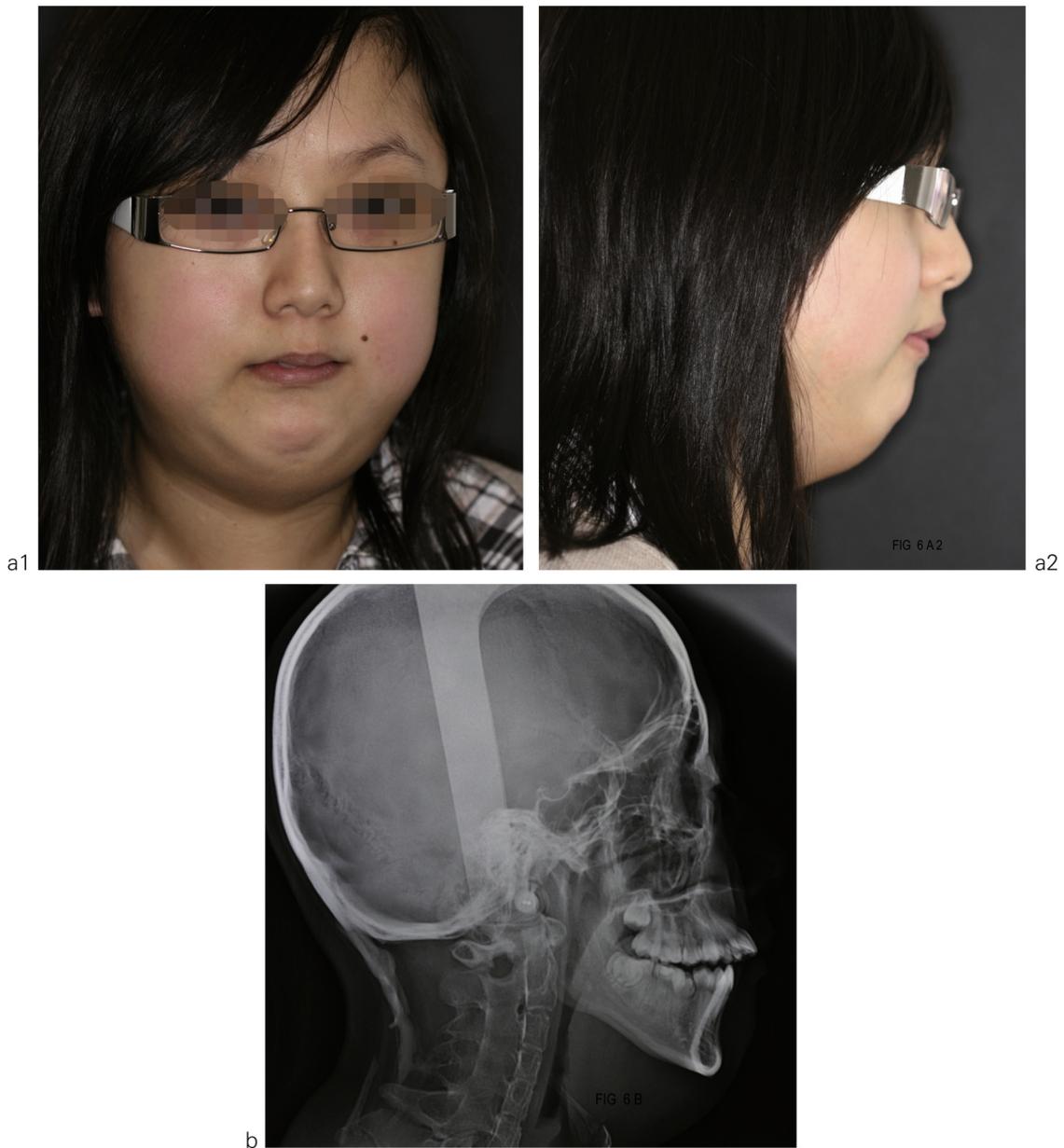
b: Preoperative panoramic xray after maxillo-mandibular osteotomy and right subapical osteotomy for reducing the height.

d: Postoperative facial and occlusal views.

In some cases, the bone structure has a peculiar appearance. For example, in patients with osteogenesis imperfecta we find some dysmorphism. In these cases, performing an osteotomy is safe including bone consolidation for very “fibrous” bone that might make the surgeon apprehensive about the difficulties involved.

### Relapses

The major difficulty for treatment plans in these areas is still the relapses that can occur because intrinsic abnormalities are not “cured” by orthognathic surgery. The surgery will make it possible to normalize osseous and occlusal structures, but the



*Figure 6*

*Female patient with progressive ossifying myositis for which surgical procedures are prohibited given the risk of seeing muscular ossification worsen.*

*a 1 and 2: Facial and lateral views*

*b: Lateral cephalometric xray showing significant calcification of the posterior cervical muscles and all of the spinous and transverse processes of the cervical spine.*

physical abnormality remains. Therefore, even though myopathies are treated properly, they often lead to a deterioration of the result (Fig. 7). In other cases they are anomalies located in one or more functional matrices that are the cause of the deterioration of the result. However, these pathologies are less likely to relapse than when the whole aggregate of mesenchymal tissue is affected. An example of this type of case can be found in deteriorations encountered secondary to facial asymmetries in hemifacial microsomias (Fig. 8). Even if, concerning this last example, the deteriorations are more significant and develop more slowly. Finally, these deteriorations of results should not deter the practitioner from providing treatment since, in a great number of cases, the results can be stable and can transform the daily life of these patients. The deteriorations may in the end be quite limited when the therapeutic treatment plan is carefully followed (Fig. 9) or when the affliction is moderate (Fig. 10).

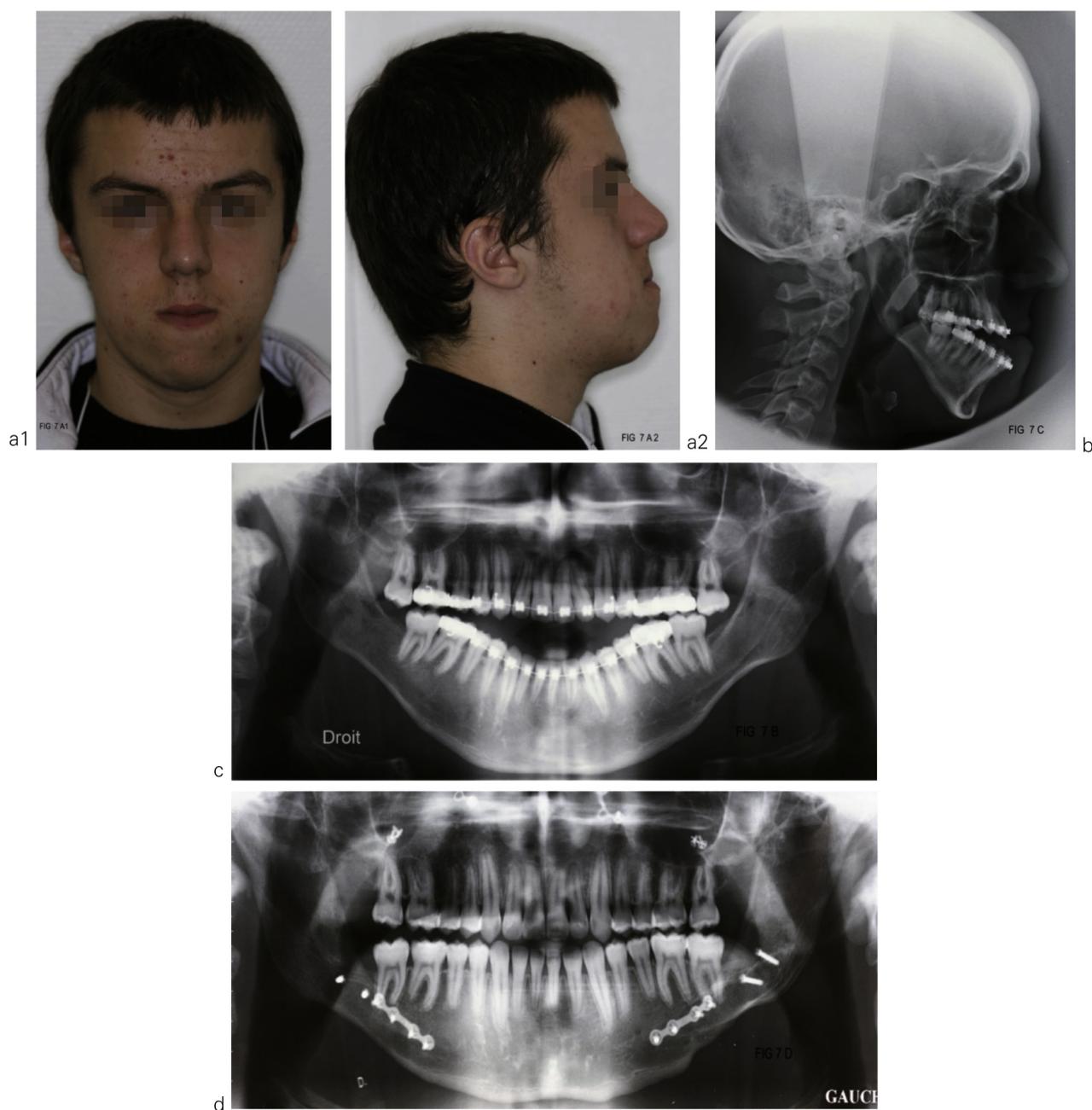
### **The compliance of patient with this type of treatment plan**

The above is a major factor in the management of the treatment plan of these patients. Although the surgical procedure itself is relatively simple, patient compliance with the treatment is sometimes lacking. This failure can cause postoperative com-

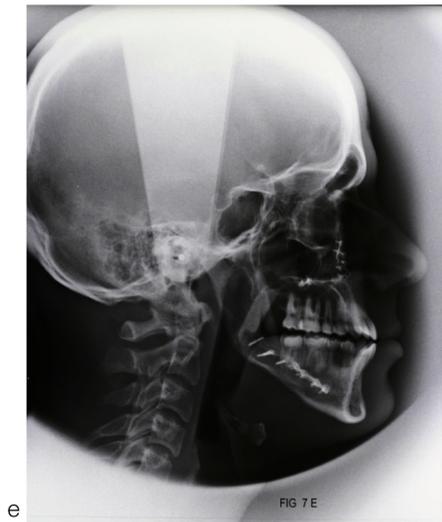
plications or partial results. In fact, it is mandatory that patients strictly follow postoperative instructions in these areas more than elsewhere so as to ensure proper occlusion by adhering to the treatment plan. In these cases, intermaxillary fixation may be extensive and can vary in how long it lasts. It should be pointed out that intermittent intermaxillary fixation is often more difficult to ensure than a permanent one maintained during the period of consolidation. Quite obviously, it is a matter of judgment and we cannot sufficiently stress the need for the patient, the institution as well as the family to actively collaborate during the treatment of these cases.

### **The follow-up treatment of these patients**

This point is particularly important given the high risk of relapse. Postsurgical occlusal stabilization in this instance more than others is of the utmost importance to have the greatest possibility of achieving a stable occlusion. Unlike treatments for more common areas where deteriorations of results occur during the first year, the relapse may continue well beyond this period due to the persistence of physical anomalies or dysfunction. Retainers are recommended and the practitioner must make sure that they do not negatively impact oral-dental hygiene.



*Figure 7*  
 Patient presenting an unidentified myopathy.  
 a 1 and 2: Facial and lateral views: Note the extreme divergence, Class III.  
 b: preoperative lateral cephalometric xray.  
 c: preoperative panoramic dental xray.  
 d: postoperative panoramic dental xray.



e



f1



f2

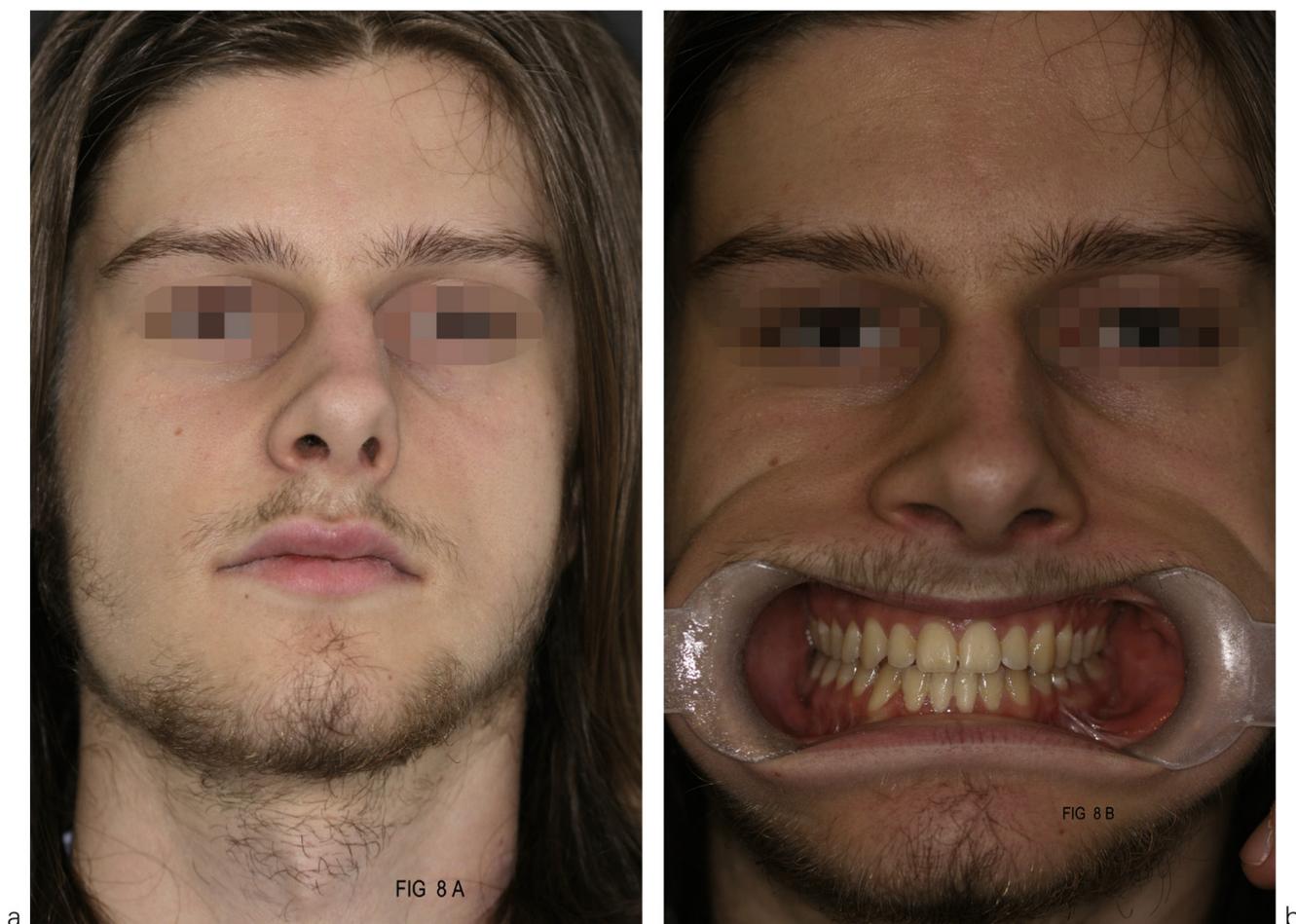


g



h

- e: Postoperative cephalometric xray, notice the deterioration of results due to muscular dysfunction.  
 f 1 and 2: Postoperative facial and lateral views.  
 Notice the tendency towards deterioration of results.  
 g: Preoperative occlusion. Notice the open bite.  
 h: Postoperative occlusion. Notice the tendency towards deterioration of results.*

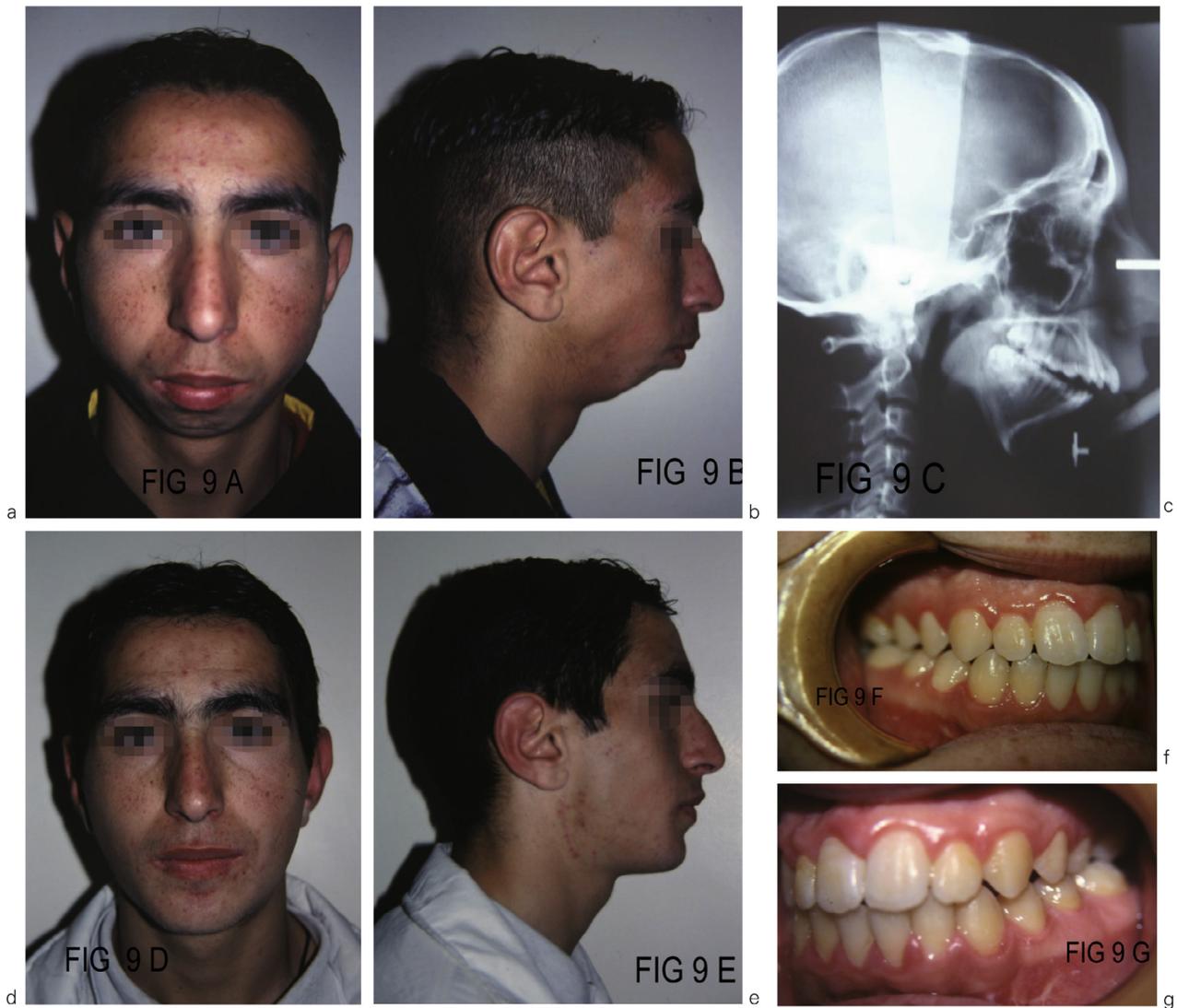


*Figure 8*

*Patient presenting deterioration of results of the hemifacial microsomia.*

*a: Clinical appearance of the patient's face showing a persistent left flattening that occurred long after orthognathic surgery.*

*b: Front view showing that occlusion is shift to the left.*



*Figure 9*

*Results of a significant condylar dysplasia treated solely by mandibular osteotomy and reconstruction of the ascending rami using costochondral graft.*

- a: Preoperative facial view.*
- b: Preoperative lateral view.*
- c: Preoperative lateral cephalographic xray.*
- d: 2 yr postoperative facial view.*
- e: 2yr postoperative lateral view.*
- f: Postoperative right lateral view.*
- g: Postoperative left lateral view.*



*Figure 10*  
*Patient with Steinert's Disease.*  
*a: Preoperative lateral view.*  
*b: Preoperative lateral cephalometric xray.*  
*c: Postoperative profile view.*  
*d: Postoperative lateral xray.*  
*e: Postoperative right occlusion.*  
*f: Postoperative left occlusion.*

## 5 – CONCLUSION

Over time, orthognathic surgery for rare diseases has increasingly been performed due to the growing concern of patients and families about dental problems. The oversight of surgery and orthodontic treatments has contributed to the use of this surgery even though as in in cases discussed above, the procedures are rarely standardized.

Generally speaking, surgical procedures and orthodontic treatment are

not one-size-fits-all techniques and because of the “areas” treated in these cases, the procedures are often more complicated.

We also must accept the inevitable fact that in these cases there is a risk of relapse due to the underlying pathology. Nonetheless, we should offer orthosurgical treatment when a request is made but there must be at least the minimum of patient cooperation.

Conflicts of interest: none

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