Developmental Anomalies of the Temporomandibular Joint

R. Bruce Ross, DDS, MSc, FRCD
Division of Orthodontics
The Hospital for Sick Children
Toronto, Ontario, Canada

Professor
Faculty of Dentistry
University of Toronto
Toronto, Ontario, Canada

Correspondence to:
Dr R. B. Ross
Faculty of Dentistry
University of Toronto
124 Edward Street
Toronto, Ontario
M5G 1G6 Canada
Fax: 416-813-6375
E-mail: bruce.ross@sickkids.on.ca

The processes by which the human face develops in the embryo are exceedingly complex, but they work out perfectly—almost every time. Occasionally, however, the development of structures such as those comprising the temporomandibular articulation is disturbed, leading to an anomalous morphology in later life. It is important to note that an anomaly is not necessarily an undesirable condition requiring treatment. It may be benign, with no associated problems, and therefore of no consequence; in fact, it may never be identified. Ross and Johnston[1] have published a review of the developmental processes and etiology of undesirable conditions affecting the temporomandibular joint (TMJ).

Craniofacial anomalies may be caused by genetic faults (eg, Treacher Collins syndrome) or caused by a teratogenic agent (eg, thalidomide), but most often it appears the cause is multifactorial (eg, cleft lip and palate). When the entire human genome is available and studied in persons with congenital anomalies, there may well be indicators for, or at least a demonstrable predisposition to, developmental problems.

Problems involving the TMJ can be acquired or congenital (developmental). The vast majority seen in a dental office are acquired dysfunctions, traumatic injuries, or pathologic conditions. Developmental anomalies appear to be rare because most of them are asymptomatic and never come to the patient's or dentist's attention. Others are associated with syndromes, and the patients are referred to centers where treatment is provided by multidisciplinary teams of specialists.

Diagnosis

The standard approach to any suspected morphologic or functional anomaly of the temporomandibular articulation is a radiograph or series of radiographs, which provides an excellent image of the bony elements. Dysmorphologies of the condyle and fossa are frequently detected in panoramic films that are part of a general examination of "normal" patients. There is thus much experience and information available on the skeletal elements.

The soft tissue elements are not as well documented. The enormous variety of developmental anomalies that occur in the craniofacial region suggests that anomalies occur in virtually every

Key words: temporomandibular joint, developmental anomalies, hemifacial microsomia

J OROFAC PAIN 1999;13:262–272.

component of every structure, even if most go undetected. If this is true, mild and even severe anomalies of the soft tissue components of the TMJ undoubtedly do occur, producing symptoms that are difficult to diagnose. Assessment of joint dysfunctions or dysplasias is usually limited to the interpretation of radiographs, clinical observations, and occasional arthroscopic examination. We do not routinely search with other sophisticated imaging techniques. Abnormalities of the synovial membrane, the capsule, and the supporting ligaments may not produce recognizable primary symptoms but may lead to secondary bony symptoms. Absence of the capsule, for example, if it occurs, would probably remain undiagnosed until it resulted in inflammation and bony destruction or ankylosis of the joint.

Developmental bony ankylosis is very rare, if it occurs at all. A decreased mobility of the mandible is most probably caused by the soft tissue elements of the joint. In an unpublished survey of 107 children with hemifacial microsomia, we found only 2 patients who had a severe restriction of opening (less than 15 mm). If developmental processes cause a dysmorphology of the bony elements, they will almost certainly cause dysmorphology of the associated soft tissues. Surgeons operating in the region of a dysmorphic condyle often note gross defects of the disc and supporting structures.

One would suspect that there are developmental defects limited to the soft tissues, but this is speculation, because access to study such joints is rare. However, disc displacement problems in adults could be a developmental malformation of the capsule and ligaments, or abnormal disc morphology. When the joint is not functioning well in a congenital problem, in my experience the bone is always recognizably involved. There may or may not be involvement of the muscles and soft tissues.

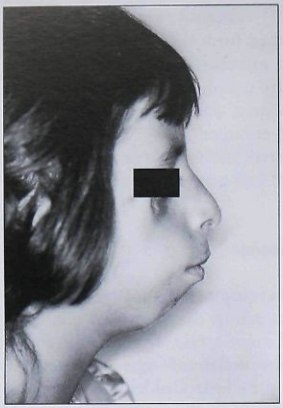
Micrognathia

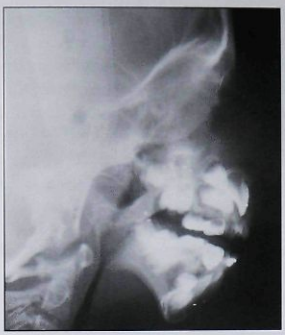
The mandible in any of the recognized micrognathias invariably has a normal appearance of the condyle and a normally functioning articulation. There is a posterior displacement of the condyle in mandibulofacial dysostosis (Treacher Collins syndrome), which may be responsible for the limited mobility and unusual shape of the lower border with time (Fig 1). True condylar hyperplasia with enlargement of the condyle does occur but has only been seen in our clinic in cases of unilateral condylar hyperplasia, a condition of unknown etiology that occurs in young adults between approximately 19 and 23 years of age. Despite the dysmorphology, TMJ function is not abnormal in these cases. Another form of unilateral hypertrophy is part of a generalized hemihypertrophy involving the entire mandible and the teeth, many other facial structures, and even the entire body. The TMJ on the enlarged side may be well forward of the external auditory canal. The affected condyle is longer but not increased in diameter (Fig 2).

Hemifacial Microsomia

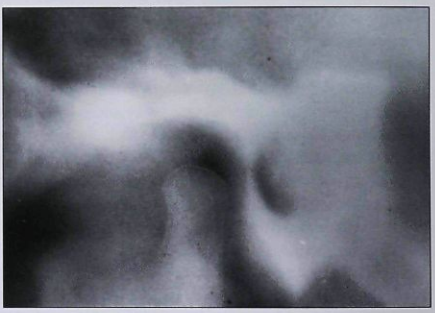
Almost all the developmental anomalies of the TMJ belong to a group of anomalies generally termed *hemifacial microsomia,* although the conditions are not always unilateral—nor are they often microsomia. They include Goldenhar syndrome and other variations.[1,2] In the severe expression of this condition there is a dysmorphology of most of the structures in the region, including the mandibular condyle and ramus, the middle and external ear, orbit, zygoma, and maxilla (Fig 3). The affected condyle and ramus are often severely dysplastic or even absent or displaced medially; in very severe cases there may be no temporal fossae or middle ear (Fig 4). The muscles of mastication are severely hypoplastic or may be virtually absent. The abnormalities of the TMJ range from complete agenesis to subtle differences in form or size with few deformities (Fig 5). Invariably, however, the dysplasia is both a deficiency and a malformation. A significant feature is that in 70% of cases the condition appears to be unilateral, while in bilateral cases asymmetry is the rule, with only subtle dysmorphology on the less affected side. Rarely are both sides severely affected. Although there is evidence of a genetic etiology in a few cases, it is not a major factor.

Surprisingly, there are almost no problems associated with the grossly abnormal temporomandibular articulation in these cases. There is no dysfunction, in the general sense of function. These patients have no associated pain or discomfort, can chew and swallow comfortably and well, can speak with normal articulation and nasal resonance, and can breathe adequately (the mandible provides support for maintenance of the airway, so that obstructive sleep apnea is very rare). There is no dysfunction except in the narrowest of definitions—that is, the mandible and TMJ do not function in the normal way. There is often a deviation of the mandible toward the affected side on opening, indicating a restriction of movement in the affected joint. This is probably due to an absence or virtual absence of the lateral pterygoid muscle





Figs 1a and 1b Child with typical Treacher Collins syndrome.



Figs 1c and 1d Two affected individuals in whom the condyle appears to be adhering to the posterior of the glenoid fossa. This may be an abnormality of the discomalleolar ligament characteristic of this condition.

or to hypoplasia of contiguous soft tissues. The affected mandible can often be freely advanced manually (Fig 6).

There are, however, psychosocial problems related to the facial asymmetry that is invariably present. One side of the mandible is greatly reduced, usually in all 3 dimensions: vertical height, anteroposterior length, and bicondylar/bigonial width. Although poor facial esthetics are a serious problem for the patient and the family, one could even argue that facial esthetics are not a biologic characteristic but a sociologic one, and in a society that did not care about physiognomy, there would be no problem with having this facies.

Timing of Treatment

When should treatment be instituted in these cases? The parents would like treatment completed in the neonatal period, if that were possible. The speech pathologist would like the speech mechanism corrected in the first year, the psychologist would like the facial appearance treated before school age at least, the orthodontist would like a corrected jaw relationship stabilized in the primary dentition, and the surgeon might like to wait as long as possible to avoid facial changes with growth after surgery. As an example, the surgical protocols for cleft lip and palate usually are: lip

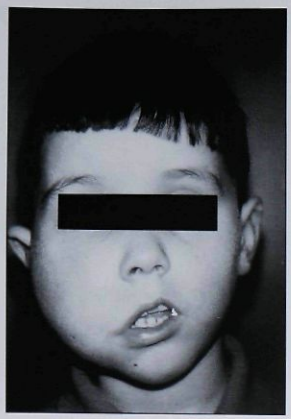


Fig 2a Child with typical hemifacial hypertrophy, featuring gross overdevelopment of many structures of the right side of the face, including the ear and the teeth.

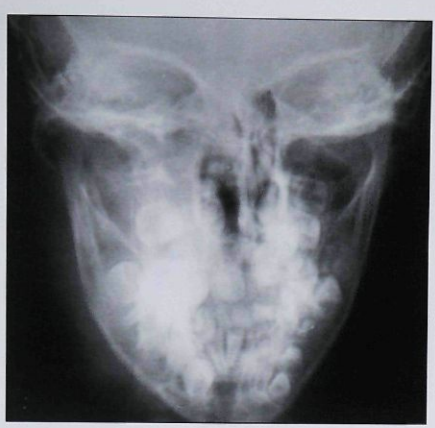


Fig 2b Radiographic view. The soft tissues are more affected than the skeleton.

Figs 2c and 2d The right condyle (*left*) is tubular in shape and the TMJ is well anterior to the external auditory canal, compared to the unaffected side (*right*).

repair in the early postnatal period for esthetics, hard and soft palate repair later in the first year for speech, alveolar cleft bone grafting at 9 to 11 years, and orthognathic surgery, if necessary, delayed until 15 to 20 years, when facial growth will not be a complicating factor.

There are some circumstances in which treatment timing is dictated by the condition and the expected changes with time. If a condition threatens the life or health of the patient, for example with obstructive sleep apnea related to micrognathia, then treatment must be instituted immediately. If the condition will gradually worsen if left untreated, for example with mandibular ankylosis, then early intervention is indicated. If the condition poses no serious problem and is stable, we have the luxury of treatment when it is most convenient for all concerned: the patient, the parents, and the health care professionals.

We have long contended that the facial and bony asymmetries in hemifacial microsomia are invariably stable during growth of the child[3,4]: a few worsen slightly, and a few improve without treatment. Careful tracings of the lateral and

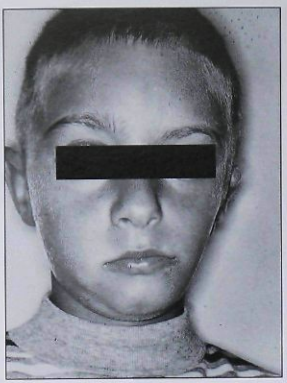


Fig 3a Child with hemifacial microsomia, showing facial asymmetry.

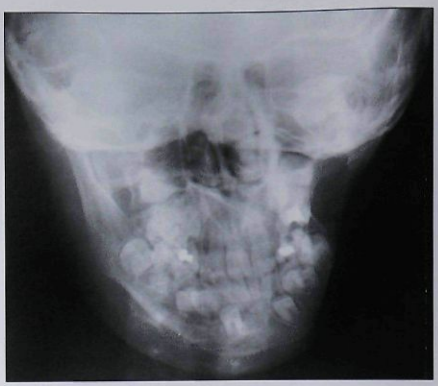


Fig 3b Radiograph showing skeletal asymmetry.

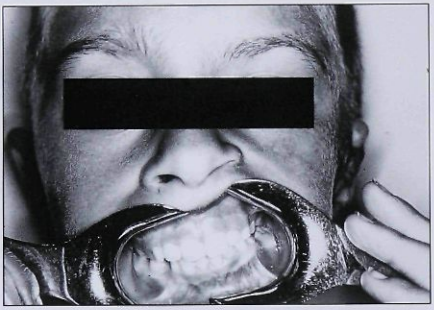
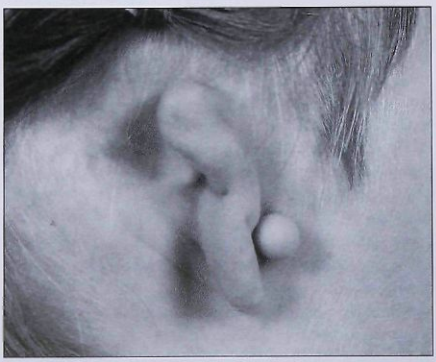
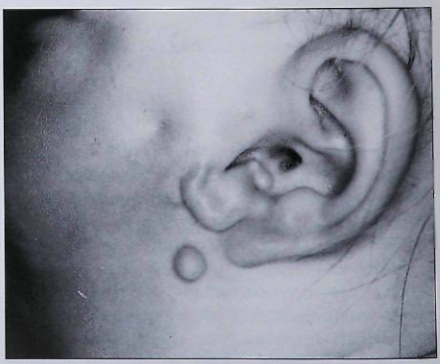


Fig 3c Canting of the occlusal planes can be seen in both the maxilla and mandible relative to the orbital plane.





Figs 3d and 3e Microtia may take the form of severe deficiency of the external ear or excessive ear tissue with preauricular tags.

Figs 4a to 4f Examples of dysplasia of the temporal bone and abnormalities of the posterior of the glenoid fossa in children with hemifacial microsomia. In every case, however, the mandibular condyle is reasonably normal and the joint functions acceptably.

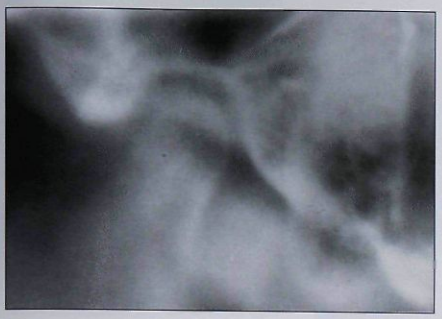


Fig 4a

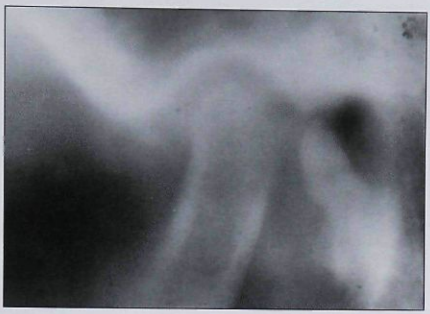


Fig 4b

Fig 4c

Fig 4d

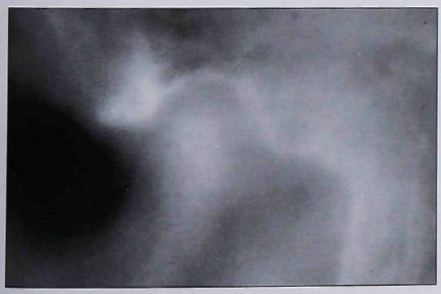


Fig 4e

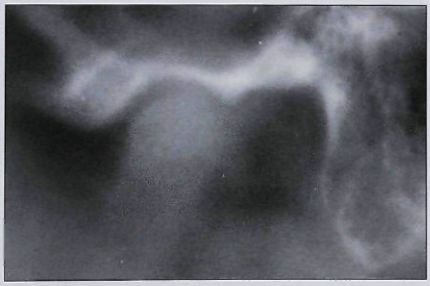
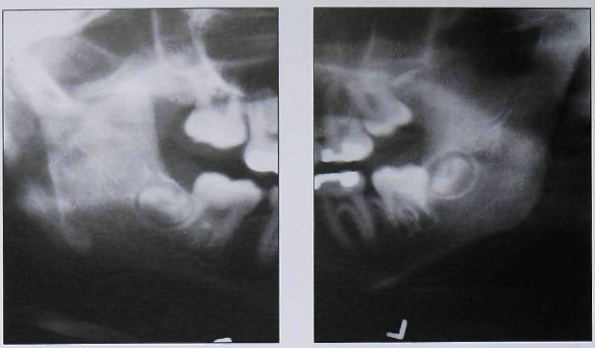
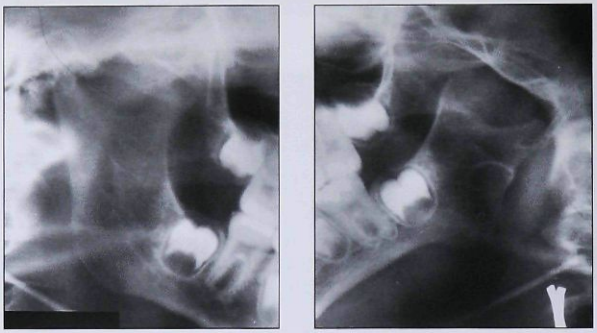


Fig 4f



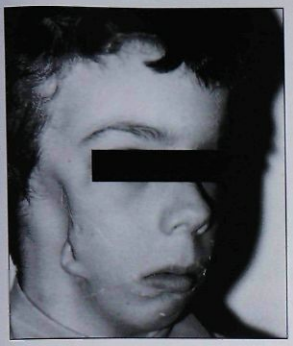
Figs 5a and 5b Right and left sections of a panoramic radiograph showing a moderate dysplasia of the right condyle and ramus.

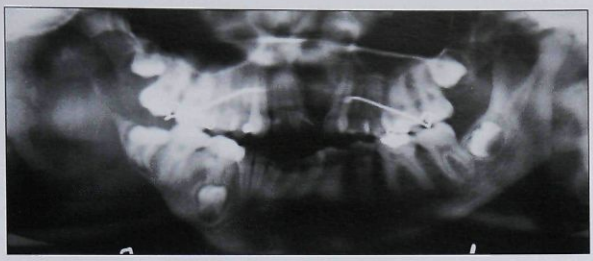


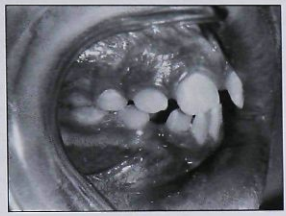
Figs 5c and 5d A more severe dysplasia of the left ramus (*right*), with no TMJ present. Highly variable irregularities of form and structure occur in these cases. The muscle attachment size (gonion, coronoid process) reflects the functional level and bulk of the muscles.

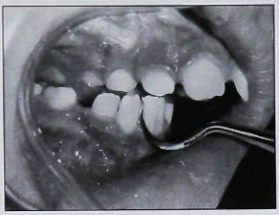
anteroposterior cephalometric radiographs and panoramic films indicate that growth of the dysplastic ramus and condyle is quite profuse and continues at or near the growth of the contralateral "normal" side (Fig 7). The existing asymmetry is maintained through childhood, showing no clinical sign of change. The orbits and maxilla also do not change perceptibly in these patients, and the tilt of the occlusal and mandibular planes does not alter. In infants, fat pads in the cheeks may disguise the skeletal asymmetry. What sometimes seems to be the appearance or worsening of asymmetry with growth may be an illusion attributed both to the thinning of the soft tissues and to the increase in the height and depth of the lower face with growth, making an existing asymmetry increasingly obvious. Occasionally there is a dramatic regeneration of a severely dysplastic ramus/condyle that is difficult to explain.[1] Polley et al[5] showed conclusively in a long-term longitudinal study that hemifacial microsomia is not progressive, and that growth of the affected side parallels that of the non-affected side almost exactly, regardless of the degree of the initial deformity. There is an impression given in the literature, however, that these cases worsen with growth. Harvold et al,[6] Mulliken et al,[7,8] and Murray et al,[9] among others, have emphatically claimed a progressive deficiency, but presented no data to support their contention. Unfortunately, their opinions have been widely repeated as a proven hypothesis, and the "worsening" misconception has greatly influenced treatment methods.

In moderate cases of hemifacial microsomia, the timing of treatment depends on the attitude of the patient and parents and is a judgment call. If they are not overly concerned with the facial difference, and it does not appear to be causing a psychosocial problem, then it is probably preferable to wait until facial growth has neared completion and the patient enters adolescence, when social activities may cause more concern about facial differences.

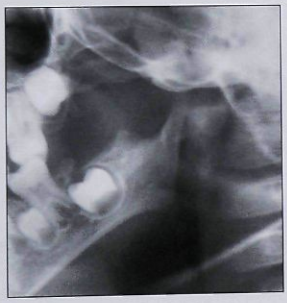


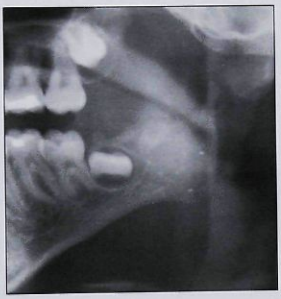






Figs 6a to 6d This patient has virtually no mandible on the right side posterior to the premolar region except for the teeth and alveolar bone. In spite of this, an excellent position of the mandibular midline is maintained rather precariously by the musculature: a gentle force can displace the right mandible posteriorly several centimeters.





Figs 7a and 7b Profuse growth of the severely dysplastic ramus and condyle does occur. Comparison of radiographs at age 9 (*left*) and age 15 (*right*) shows that approximately 17 mm of bone was deposited on this ramus in 6 years, which provided space for development of the third molar. This was only 2 to 3 mm less than on the contralateral side. The stimulus for growth was not function, since there is no TMJ on the left side.

Treatment

If the TMJ is dysmorphic, leading to esthetic concerns in the patient or parents, then a decision must be made as to whether improving the facial esthetics requires surgical reconstruction of the temporomandibular articulation, mandibular lengthening without interfering with the joint, or soft tissue augmentation to build out the deficient side of the face. If the TMJ is dysmorphic but functioning and asymptomatic, it is best to leave it as is and treat the asymmetry by sagittal split osteotomy, distraction osteogenesis, or soft tissue augmentation; there is no valid reason for any surgery to the joint.

If the TMJ is dysmorphic without reliable condylar interaction with the temporal bone (ie, no real joint), or if the ramus deficiency is severe, then a joint must be constructed and bone added to achieve mandibular symmetry and acceptable facial esthetics. The major controversies with these patients are how to replace the missing skeletal

tissues and when to begin surgical treatment. A favored method of reconstructing the ramus and condyle and establishing a functional articulation with the temporal bone (a pseudo-joint) is by means of a bone graft to the mandible (most commonly a costochondral graft). The costochondral graft will function well as a condyle in this situation and will even remodel to the bulbous shape of a condyle. The indications for grafting are the absence of a functional joint with consequent severe facial asymmetry.

Early Surgery

The advantage of an early approach is that the child has a reasonably symmetric face throughout childhood, even though a second fine-tuning surgery might be necessary at the conclusion of growth. Treatment by costochondral grafting has been shown to be more effective if performed at an early age, about 4 to 5 years.[10] At this age the success rate is higher, the dentition adapts spontaneously to the corrected arch relationship, and growth of the costochondral graft keeps pace with the growth of the "normal" side in most patients. The orthodontist would prefer correction as early as possible to establish a symmetric mandible. The erupting permanent teeth will then assume a more normal relationship to the underlying basal bone and the face in general, and not be forced by the abnormal matrix to develop severe compensatory positions that are difficult to reverse later. Restoration of more normal tissue forces early in growth will permit a relatively simple finishing procedure at the conclusion of facial growth in adolescence. Early reconstruction is essential to the management of ankylosed joints as well.

Surgery at 3 to 5 years of age will alleviate the impact of a severe facial deformity on the child during the early school years, when self-esteem is fragile and patterns of social interactions are developing. Development of the dentition is better if the jaw relationship is close to normal at an early age. It would appear that early TMJ construction by costochondral grafting is, at present, the method of choice for severe hemifacial microsomia. This appears to be a very useful surgery, with a failure rate that could be considered tolerable, given the alternatives. There is no other way to improve the severe cases, in our experience. Functional appliance therapy is of little value in severe cases, and distraction osteogenesis of the mandible is not indicated in cases where no TMJ is present.

Late Surgery

The alternate approach of delaying surgery until adolescence has the advantage of more stable structures, eliminating growth as a factor. The expectation is that only a single surgery will be necessary, although experience indicates that a second finishing surgery is often required. The great disadvantage of this approach is that the child must live with a facial deformity throughout childhood.

Early surgery, however, carries the potential problem that the graft and the opposite "normal" side will have different growth rates. Lesser growth of the graft may be related to operative or postoperative complications, rather than to a direct growth phenomenon. A serious problem when it occurs is that of extravagant overgrowth of the graft.

A possible explanation for growth differences may lie in the size of the germinative zone of cartilage in the graft. The prechondrocytes in this zone supply cells for the proliferative zone, where interstital growth is responsible for increased length of the cartilage. Peltomaki and Ronning[11] have shown that when costochondral grafts were transplanted to a non-functional area in rats, the growth in length of the graft varied with the thickness of this zone of cells. Removal or injury to these cells inhibited growth. If these findings can be extrapolated to the human condition, it may be possible to achieve clinical control of the postoperative growth of the costochondral graft by adjusting the amount of cartilage available. The reduction in growth in cases reported by Perrott et al[12] may have resulted from excessive trimming of the cartilage, while the many noted examples of excessive growth may have been associated with inadequate trimming. Peltomaki and Ronning[13] also showed that mature, non-growing ribs transferred to a non-functional area in growing rats grew significantly. Their findings indicate a systemic, hormonal stimulation rather than a functional one.

The creation of a symmetric face is generally possible only with reconstructive surgery. Orthodontics is unsuccessful in all but the mildest cases, although it is an important adjunct to the preparation and retention of the surgical plan. In almost every case, some form of surgical treatment is required to achieve a satisfactory result. Orthodontic treatment provides the beautiful smile that contributes so much to overall facial esthetics. However, some orthodontists have believed the "worsening with growth" theory

discussed above and believe that they influence growth with functional orthodontic appliances. In the opinion of Harvold and his supporters,[6] growth of the affected condyle while a functional appliance is being worn proves the effectiveness of the appliance, when in fact, profuse growth would occur without appliance intervention. They admit that in severe cases "effective length increase of the mandible cannot be achieved by treatment with a functional appliance" and that even considering mild cases "it is rare that surgical procedures on the jaws . . . can be avoided."[14] There are scattered anecdotal accounts of great improvement, but the data are questionable.

Functional Appliances

Treatment involving extended use of functional appliances in the expectation of growing an adequate mandible seems to many clinicians to be a futile and wasteful procedure. In patients with normal mandibles, there may be an increase in mandibular length of several millimeters with the use of a functional appliance. It is unlikely that this would be exceeded in severely dysmorphic mandibles, and that amount of correction would be grossly inadequate in patients with hemifacial microsomia. Functional appliance wear probably constitutes a severe traumatic experience for these particular patients, poses significant logistical problems, and represents an appreciable expense. In many cases it requires a delay of surgery well beyond the age that we consider preferable. In our experience these appliances are unsuccessful and contraindicated in all but the mildest cases, where the occlusal plane can be manipulated and teeth aligned to an extent that may obviate some or all of the need for surgery.

The proponents of functional appliances further insist that, following mandibular surgery, continuous wearing of functional appliances is essential during the remaining growth period to induce further growth of bone.[14] No appliances were used in any of our patients who were growing, however, and growth of the new condyle was profuse.

The technique of distraction osteogenesis is becoming popular as a means of lengthening one or both sides of the mandible. This technique appears to have many advantages, but it requires an existing functional articulation of the condyle with the temporal bone to be successful. Frequently, there is no such articulation in severe cases of hemifacial microsomia.

Differential Diagnosis

It is useful and often essential to determine whether a malformed mandible or facial asymmetry is developmental or acquired. There are many indicators that make this an easy diagnosis, as outlined below.

- Pain indicates an acquired anomaly. There is no pain or discomfort with developmental anomalies, and no history of pain.
- A change over time in the condition, either the appearance of new symptoms or a change in appearance, indicates an acquired anomaly.
- A temporary change in symptoms associated with function (eg, heavy chewing) indicates an acquired anomaly. Developmental anomalies are not affected by function.
- Abnormal size or function of the facial muscles suggests a developmental etiology (eg, deviating soft palate, hypoplasia of masseter or temporalis).
- The presence of other developmental anomalies of contiguous structures (eg, ear, eye, palate, macrostomia) suggests a developmental etiology of the TMJ problem.
- Crepitus or clicking in the joint indicates an acquired anomaly.
- Ankylosis is almost always, if not always, acquired. Limitation of mandibular forward translation may be developmental, but rotation is not usually limited to any great extent. Hypermobility is usually congenital.
- Marked dental compensations for an asymmetric mandible indicate a very early development of the asymmetry, possibly congenital or in infancy. The corollary to this is that if there are severe crossbites, the condition was probably acquired at a later time.
- The condyle in a developmental condition may be deficient or excessive, but it is round and smooth. If the condylar head is flat or an irregular shape, it is an acquired anomaly.

Conclusions

Congenital anomalies of the temporomandibular articulation usually function satisfactorily. There is no discomfort with developmental anomalies, even with severe joint dysmorphology. Furthermore, the joint itself requires little structure to function satisfactorily. Functional lateral or protrusive excursions of the mandible are often noted in individuals with developmental anomalies but also do not appear to result in TMJ symptoms.

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