Chapter 67: Noncleft Disorders of the Oral Cavity and Oropharynx

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The noncleft congenital anomalies of the oral cavity and the oropharynx are many and diverse. Congenital anomalies are those present at birth regardless of the cause, whereas genetic anomalies need not be present at birth but become detectable later. Thus congenital anomalies can be secondary to genetic, related abnormalities or to isolated abnormalities secondary to embryologic defects of unknown or drug-related origin. Consequently, one can divide congenital anomalies into those that are hereditary and those that are nonhereditary. The other mode of classification is to separate isolated anomalies of the oral cavity and oropharynx from syndromes that involve the oral cavity and oropharynx.

For accurate assessment, correct diagnosis, and management, the many problems children with disorders have should be dealt with in a team approach. Such teams, which exist in major centers dealing with craniofacial anomalies, include a pediatrician, neurologist, geneticist, otolaryngologist, plastic surgeon, dental specialist, prosthodontist, orthodontist, oral surgeon, social worker, psychologist, audiologist, and speech therapist.

Assessment

Minimal assessment should include a thorough physical examination, including a family history, radiologic studies of the skull and face, chromosomal studies, and any other documentary procedures (such as videotaping and fideofluoroscopy). Because these patients have speech problems, such as velopharyngeal insufficiency, a speech therapist should perform an adequate speech assessment. Following this, an examination with a flexible nasopharyngoscope and documentation by videotaping and multiview videofluoroscopic speech studies are mandatory before the necessary treatment is begun.

Although feeding is not as severe a problem for parents as it is in patients with cleft abnormalities, it can be a bothersome procedure, especially in patients with a Pierre Robin sequence or nasal obstruction secondary to remnants of the buccopharyngeal membrane.

Nutrition should never be decreased for any reason, even if maintaining it requires a long-term indwelling nasogastric tube. Such a tube has many complications, especially erosion of the posterior cricoid and gastroesophageal reflux. Gastrostomy is a good alternative.

Upper airway obstruction can be life threatening and is hazardous to the cardiac and pulmonary status of the patient (cor pulmonale). Upper airway obstruction should be vigorously treated. With oral cavity and oropharynx abnormalities, the obstruction can be secondary to macroglossia, which can be found in Down syndrome, but rarely necessitates major intervention; or it can be secondary to hypognathism, such as in Pierre Robin sequence. Congenital lingual cysts have been reported to cause respiratory distress. Craniofacial dysostosis from Apert-Crouzon syndrome can reduce the size of the nasopharyngeal airway and lead to cor pulmonale (Pruzansky, 1973) much as an enlarged adenoid can do. In cases that are very difficult to treat, an adequately observed tracheotomy can be a safe procedure.

Ear disease is mainly secondary to abnormalities in eustachian tube function, which in most of the cases is caused by mispositioning of the eustachian tube and partial malfunction of the tenros veli palatini and levator palatini muscles. Most of the children with craniofacial anomalies, of which the oral cavity and the oropharynx manifestations is a part, have effusion problems with otitis media. Such patients should be treated vigorously, usually with tympanostomy tubes, and then monitored very careully because of the chronic complications (such as hearing loss) that can ensue from recurrent ear infections.

Poor dental occlusion, in addition to creating feeding, speech, and cosmetic problems, can be associated with severe temporomandibular joint problems and chronic pain (Costen's syndrome), which has been overemphasized in the old literature. Prompt diagnosis and management by the dental team are mandatory in such cases. Further, because of cosmetic deformities that can be present from abnormalities in the positioning of the maxilla and mandible, major surgery can be anticipated (mandibular and maxillary advancement or setback). Poor oral hygiene, sometimes caused by the disease process but more commonly by neglect, usually exists, causing more problems. Drooling can be a bothersome manifestation, especially in patients who have associated neurologic abnormalities. Drooling is treated surgically by excising both submandibular glands and tying Stenson's duct on one or both sides (Shott et al, 1989).

The patient's problems should be monitored carefully. Family members and friends should give every support to help achieve the goal of a good correction with the patient's cooperation and to encourage the patient to develop an independent, active personality.

Specific Localized Anatomic Anomalies

The oral mucosa, gingiva, lips, tongue, maxilla, mandible, and teeth compose the oral cavity and oropharynx. This chapter discusses abnormalities that involve each of these structures alone, taking into consideration that some tumors appear at birth and can be anywhere in the oral cavity or the oropharynx. Such tumors include hemangiomas, hamartomas, cystic hygromas, craniopharyngiomas, chordomas, teratomas, neuroblastomas, gliomas, Thornwaldt's cysts, heterotopic brain tissue, and botryoid rhabdomyosarcomas. The management of these tumors is the same as that for tumors in any other situation; one should keep in mind, however, that newborns are medically fragile, and thus every efort to maintain feeding and respiration and to prevent infection should be made.

Anomalies of oral mucosa

Hereditary benign intraepithelial dyskeratosis (Witkop's disease) is an asymptomatic lesion manifested by a white- or cream-colored-plaque that can involve the tongue and floor of the mouth. This benign disease is secondary to dyskeratosis of the oral mucosa.

White sponge nevus is an asymptomatic innocuous condition manifested by white spongy plaques of the oral mucosa. It is not a premalignant or malignant lesion. The condition is transmitted as an autosomal dominant trait.

Hereditary mucoepithelial dysplasia, transmitted as an autosomal dominant trait, is a red and flat micropapillary lesion that is usually found on the gingiva and palate but can involve all the oral mucosa.

Fordyce (1896) was the first to describe buccal and labial sebaceous glands. Fordyce's spots are small, slightly elevated to flat, yellowish brown spots occurring singularly or in groups on the oral mucous membranes and vermilion portions of the lips. Incidence has been variable. Schumacher reported an incidence of 30%, whereas other reports (Halperin et al, 1953) showed and 82% incidence in a total population of 2478 individuals. The incidence in males was similar to that in females, and whites were similar to non-whites. These atopic sebaceous glands can occur in many mucocutaneous sites, including the cervix uteri (Watson and Cochran, 1969). The morphology is the same as in sebaceous glands in other sites. They develop from aberrant ectodermal buds on fetal lines of closure. Many of these glands have ducts leading to the surface (Chambers, 1928). Fordyce's spots have no significance except for their close association with some rheumatic diseases, especially Reiter's syndrome (Vilppula et al, 1983).

Anomalies of gingiva and teeth

Gingival fibromatosis is a condition that is associated with many syndromes and is usually transmitted as an autosomal dominant trait. This firm, pink, painless enlargement of the gingiva is slowly progressive and nonhemorrhagic; it can be unilateral or bilateral, localized or generalized.

Mucolipidosis II is known as I-cell, or inclusion cell, disease. This gingival enlargement may impair mouth closure. Eruption of teeth is delayed because the teeth may be included in the hypertrophic gingival tissue.

Congenital epulis, a firm, nontender mass covered with pink, smooth mucosa, is usually located in the gingiva of the anterior region of the maxilla (Fig. 67-1). The mass is found on the maxilla more than on the mandible and is more common in females than in males. Congenital epulis is a relatively rare entity, with fewer than 200 cases reported in the literature (Fuhr and Krogh, 1972). Histologically, it is similar to granular cell tumors of the tongue. The theories of its origin range from odontogenic to fibroblastic to neural. Treatment is by excision, which can be done in the office. No sequelae, such as long-term damage to the teeth, occur.

Teeth anomalies are anomalies of morphology and structure; the time of eruption and number of teeth are also unusual (Fuhr and Crogh, 1972). Anomalies of morphology and structure include large bulbous crowns with an absent cusp and sulcus occlusal patterns. These can be found in otodental dysplasia syndrome. Widely spaced, flared anterior teeth and tapered posterior teeth with pointed cust tips are seen in Morquio's syndrome. Conically shaped lateral incisors or canines with barrel-shaped central incisors are seen in Ellis-van Creveld syndrome.

Anomalies of structure involve enamel and dentin hypoplasia and hypocalcified and hypomature enamel defects that are inherited and referred to as amelogenesis imperfecta. Inherited dentin defects are referred to as dentinogenesis imperfecta; the dentin is brown to

blue and wears away easily by mastication. The enamel also chips of easily because of poor physical retention at the dentinoenamel junction.

Teeth are typically not observed at birth. If they are, they are called natal teeth - usually the lower central incisors. Neonatal teeth are teeth that erupt during the first 30 days of life and are usually the lower central incisors. This condition occurs as an isolated trait (1 in 2000 or 3000) or can be associated with syndromes beyond the scope of this chapter.

Three anomalies of number may occur. Anodontia is the complete absence of primary and secondary teeth, the commonest being the third molars; 2% to 6% of the population have absence of other teeth, usually the lower second bicuspid. An autosomal dominant gene controls the upper lateral incisor. Hyperdontia, the presence of extra teeth, is less frequent than hypodontia (0.1 to 3.6%). It is usually diagnosed radiologically and is more common in the maxillary anterior and molar areas than in other areas. Both hyperdontia and hypodontia can exist as isolated traits or as parts of syndromes.

Anomalies of lips

Lip nodules are transmitted as an autosomal recessive trait and are part of pseudoxanthoma elasticum. The nodules are painless and yellowish and are found intramucosally over the lower lip and on the buccal mucosa, palate, and tonsillar areas.

Commissural lip pits are depressions at the corners of the mouth 1 to 4 mm deep and 2 to 3 mm wide. They have no clinical significance and are transmitted as an autosomal dominant trait. The incidence is 12% in whites, 20% in blacks, and 6% in Orientals (Baker, 1966).

Lip pits associated with cleft lip or palate are the surface openings of the fistulas connecting minor salivary glands to the paramedian surface of lower lip vermilion (Van der Woude, 1954). If pressure is applied, saliva can be extruded. They have no clinical significance and are transmitted as an autosomal dominant characteristic.

A lip frenulum is a small, fibrous band that connects the lower or upper lip in the midline back to the gingiva. If thick, it can partially limit lip mobility. Treatment is excision, which can be performed with the patient under local anesthesia as an office procedure.

Anomalies of maxillar and mandible

The functional and cosmetic sequelae of maxilla and mandible abnormalities are vast and devastating. Maxilla or mandible underdevelopment can cause severe upper airway obstruction with life-threatening consequences. A short soft palate secondary to underdevelopment of the maxilla causes velopharyngeal insufficiency and poor speech. The presence of maxillary cysts and embryologic fusion lines, such as globular maxillary, nasal alveolar, nasopalatine, and palatine cysts, can predispose to infection and erosion of surrounding bone. Congenital tumors in the maxilla can also be present.

Stalker and Allen (1983) have described bilateral blind epithelial-lined tracts on alveolar mucosa proximal to the inferior labial frenulum. Patent nasopalatine ducts (Farman et al, 1982) are developmental anomalies causing occasional swelling and pain with infection, nasal discharge, and squeaking or sucking. Treatment is usually surgical, but if the patient is asymptomatic, surgery is not necessary. Cohen and Abt (1970) have described the presence of heterotopic pharyngeal brain tissue. They reported a case in which a mass was broadly attached to the soft palate and right nasopharyngeal wall. They learned that brain tissue in the pharynx obstructed the upper airway and caused death of the patient. We encountered a similar patient in which a maxillectomy had to be performed at 1 month of age (Fig. 67-2).

A hypoplastic mandible can occur as an isolated entity or as part of the following first arch syndromes: Pierre Robin anomalad, Treacher Collins, cri du chat, trisomies, and Goldenhar's. Cleft of the mandible is usually secondary to maldevelopment of the first and second branchial arches and can be associated with cleft tongue and macrosomia (Fig. 67-3). Failure of the mandibular ramus to develop can be seen in hemifacial microsomia, and absence of the angle can be seen in pyknodysostosis.

Cherubism (an autosomal dominant trait) is the painless, hard, symmetric enlargement of the mandible that can start early in childhood. On radiographs, characteristic radiolucent, multilocular lesions can be seen. These maxillary and mandibular lesions eventually regress.

Torus palatinus (Fig. 67-4) is bony exostosis along the suture line of the hard palate, which can be flat, spindle, nodular, or lobular. Its incidence is approximately 21.7% in 2064 whites (Kolas et al, 1953). The incidence in American Indians and Eskimos is 60%, in Japanese it is 43.7%, and in blacks it is 16.2%. The percentage of occurrence increases after the third decade of life. It is twice as common in women as in men. The etiology can be related to functional stress, heredity, and continued growth process. It has no pathologic significance.

Torus mandibularis is bilateral or unilateral exostosis on the lingual aspect of the mandible in the region of the premolars above the mylohyoid line (Kolas et al, 1953). These tori can be single or multiple. The incidence is approximately 7% to 8% in whites and blacks; in Eskimos it is 41.8%. The incidence in men and women is equal. This incidence rises from 1.48% in the first decade to 10.6% in the third decade. Of the population, 3% have both torus mandibularis and torus palatinus; these conditions are not related. The etiology of torus mandibularis is related to functional stress and genetic background. It has no pathologic significance.

Anomalies of tongue

Tongue anomalies can be secondary to abnormal development of the tongue proper or failure of structures such as the thyroid and tuberculum impar to disappear. Tongue asymmetry, usually noted at birth when the infant has feeding problems, is the enlargement and thickening of the affected side of the tongue. Bifid tongue is a median cleft of the tongue secondary to failure of the lateral tubercles of the tongue to fuse at the midline. It can be associated with multiple lobes, usually in the anterior portion of the tongue secondary to maldevelopment of the lateral tubercles, causes lobulation of the tongue. Lingual hamartoma (Fig. 67-5) is rare; only 8 cases have been reported in the literature.

Fissured tongue (Fig. 67-6) is a congenital malformation that can be acquired; 60% of persons who reach 40 years of age have some tongue fissuring (Ray, 1963). Halperin et al (1953) found a 5% incidence, with less than 0.4% in children younger than 11 years. There is no difference in sexual or racial distribution. Vitamin B complex deficiency and repeated trauma can be the causes.

Median rhomboid glossitis (Martin and Howe, 1938) is a rhomboid reddish area in the midline of the tongue anterior to the circumvallate papillae (Fig. 67-7). The area is devoid of papillae. Incidence ranges from 5 in 2300 (McCarthy, 1941) to 8 in 2478 (Halperin et al, 1953). Embryologically it is secondary to failure of the lateral halves of the tongue to fuse before the tuberculum impar becomes interposed between them. This is not an inflammatory lesion, as might be implied by the "itis" of glossitis. The lesion is a variant of normal and should not be mistaken for a neoplasm.

Tongue-tie or ankyloglossia (Fig. 67-8) is the presence of a lingual frenulum, which can range from a mucous membrane band to a short and thick band and, in extreme cases, to fusion of the tongue to the floor of the mouth. The incidence is around 0.2% to 0.3%, but much higher (1.6%) incidences have been reported (Catlin and DeHaas, 1971). Diagnosis of this condition is by examination. The normal tongue at birth is short, so assessing tongue-tie in newborns can be difficult. With age, the tongue becomes longer and thinner at the tip (Horton et al, 1969), and evaluation for resection should include tip retriction with restraint of tongue movement so that the tip of the tongue cannot be protruded beyond the lower incisors (Wallace, 1964; Whitman and Ranknow, 1961). The effects of such a condition, in addition to speech defects and occasionally restriction of sucking, include dental deformities, such as open bite, or even prognathism. It has been suggested that treatment is usually not indicated before the age of 4 years (Watson and Cochran, 1969) unless a major sucking problem exists. In the author's experience, however, the need for division of the frenulum is usually apparent in infancy, and there is no reason to delay surgical management. Treatment is surgical. In cases of a thin mucosa a simple incision in the office is sufficient. Cases in which the frenulum is thick and the tongue adheres to the floor of the mouth may require sharp dissection, including genioglossus myotomy. Closure of the mucosa is mandatory, and many techniques, including Z-plasties, may be used. The preferred treatment is horizontal sectioning of the frenulum down to the lingual septum and then suturing of the mucosa vertically with 3-0 chromic catgut. The patient is allowed to eat after the operation.

Ankyloglossia superior (Spivack and Bennett, 1968) is a palatoglossal adhesion that is usually combined with other congenital maxillofacial anomalies, such as cleft palate and lateral synechial syndrome (Gorlin et al, 1976). Treatment is usually sectioning of the adhesion.

Lingual thyroid (Fig. 67-9) is a developmental anomaly secondary to persistence of thyroid tissue in the base of the tongue between the foramen cecum, which is immediately posterior to the V-shape formed by the circumvallate papillae and epiglottis in the midline. The whole thyroid may not descend, and around 70% of patients with lingual thyroid do not have a cervical thyroid (Myerson and Smith, 1966). Sauk (1970) and Baughman (1972) found a 10% incidence of thyroid gland tissue remnants by histologic examination of tongues in routine autopsies. Clinically the incidence is 1 per 10.000 individuals. No racial discrimination exists; symptomatic females are, however, much more common than males since thyroid

gland disturbances are more common in females and can appear at any age although they usually appear around puberty. A close association with cretinism exists.

The signs and symptoms are like those of a mass in the base of the tongue, such as dysphagia, dyspnea, and dysphonia (hot potato speech). Diagnosis is by physical examination, which shows a rounded, smooth, mucosa-covered mass of variable size in the base of the tongue. Other studies should include thyroid function tests and scintigraphy with technetium or radioactive iodine to check the presence of functional thyroid tissue in places other than the base of the tongue. Sometimes an intraoral probe may be necessary. Biopsy may be hazardous because of hemorrhage and infection (Ray, 1938).

Three treatment options are available. First, shrinking the mass can be attempted by using thyroid hormones. Second, radioactive iodine 131 may kill the thyroid tissue in the base of the tongue, but it will also kill the thyroid tissue in the neck and can be hazardous because of sloughing and possible hemorrhage. The third modality of treatment is surgical excision. The indications for surgery are dysphagia, dysphonia, dyspnea, repeared or severe hemorrhage, uncontrolled hyperthyroidism, degeneration with necrosis, and suspected malignant transformation (Ray, 1938). Excision may also be recommended prophylactically in a man over 30 years old, because, as Ray found, almost all cases of malignant transformation occur in this group. Excision can be done intraorally or through a median or lateral pharyngotomy. Autoimplantation of the ectopic gland has been suggested, but is not recommended because of the increased risk of malignant transformation.

Other congenital tumors occurring in the dorsum of the tongue include hemangiomas, cystic hygromas, fibromas, ectopic brain tissue, and hamartomas.

Anomalies of oropharynx

Both pharyngeal atresia (Morris and Reay, 1971) and congenital large pharynx (Calnan, 1971) have been reported. Patients with congenital large pharynx and velopharyngeal insufficiency are not helped with palatal push-back surgery; instead, a posterior pharyngeal flap is needed.

The presence of a subglossopalatal membrane has been reported in one girl, who developed dyspnea and dysphagia after birth (Nakajima et al, 1979). A thick, fan-shaped fibrous membrane existed from the subglossal region to the junction of the cleft of the soft palate and the hard palate. The literature records a persistent buccopharyngeal membrane (Chandra et al, 1974) and pharyngeal membrane (Hoffman, 1979) from the anterior pillar to the base of the tongue, interfering with speech, posterior pillar mucosal webbing, and palatal pharyngeal muscle displacement (Warren et al, 1978). Newcomb (1897) reported 42 cases that included absence of pillars and tonsils and lymphoid tissue abnormalities. We treated a newborn who experienced respiratory distress just after birth by excising a subglossopalatal membrane (Fig. 67-10).

Syndromes Involving Oral Cavity and Oropharynx

A Pierre Robin anomalad (Fig. 67-11) consists of cleft palate, micrognathia, and glossoptosis. The condition can be an isolated sequence or one associated with other congenital syndromes such as the Stickler syndrome, the cerebral costal mandibular syndrome, campomelic syndrome, and persistent let superior vena cava syndrome. Pathogenetically, Pierre Robin sequence is secondary to arrested development. The defect occurs when hypoplasia of the mandible prevents the normal descent of the tongue between the palatal shelves (Latham, 1966). A small mandible, which is symmetrically receded, produces an "Andy Gump" appearance.

Micrognathia offers little support of the tongue musculature, thus allowing the tongue to fall downward and backward into the lower postpharyngeal space, obstruct the epiglottis, and cause respiratory distress. Ankyloglossia is relatively common in this abnormality. The tongue size has always been controversial. Routledge (1960) suggests that cases showing delayed appearance of the anomaly are caused by disproportionate growth of the tongue during this period. Cephalometric studies (Pruzansky and Richmond, 1954) suggest that the micrognathia is insufficient to produce respiratory embarrassment unless the tongue is normal or enlarged.

Feeding problems in these children are thought to be caused by inadequate control of the tongue. Feeding problems are usually managed through specially designed nipples.

Early respiratory problems secondary to micrognathia and glossoptosis can be life threatening. In mild cases, putting the child in the prone position can help. Anterior glossopexy by passing a suture that pulls the base of the tongue anteriorly to the symphysis of the mandible has been tried. Lingual labial flaps have been suggested (Hawkins and Simpson, 1974). Our experience is that in cases in which respiratory distress exists the safest and only effective way to ensure adequate breathing is to perform a tracheotomy and follow-up of the patient. Usually decannulation occurs around 18 months after the tracheotomy and after palatoplasty.

The growth of the jaws usually progresses until a normal profile is reached by 4 to 6 years. Exceptions to this are relatively common.

Mandibulofacial dysostosis (Treacher Collins syndrome) is a syndrome that is transmitted as an autosomal dominant trait with variable expressivity. The derivatives of the first branchial arch, groove, and pouch are abnormal. These patients have nonfusion of the zygomatic arches, abnormal development of malar bones, defective orbital margins, small or absent paranasal sinuses, and congenital aural atresia with conductive deafness. Mental retardation exists in fewer than 10% (Rogers, 1964). The oral abnormalities include unilateral or bilateral macrostomia, which is present in 15%. Pharyngeal narrowing and hyperplasia are very common (Shprintzen et al, 1979) and cause respiratory complications after palatoplasty and/or pharyngoplasty for repair of cleft palate or velopharyngeal insufficiency. The mandible is hypoplastic, the ramus can be deficient, the angle is usually more obtues than normal, and the coronoid and condyloid processes are flat or even aplastic. All of these give a relative macroglossia with respiratory embarrassment, which should be managed in the same manner described or the Pierre Robin sequence.

Down syndrome (trisomy 21, mongolism) patients have a high, narrow palate and may have bifid uvula and cleft lip and palate. Either true or relative macroglossia can be present because of the small size of the oral cavity. The tongue usually protrudes, and fissuring starts to appear by 5 years of age. The middle third of the face is often hypoplastic with relative mandibular prognathism and a flattened appearance. The nasopharynx is usually narrow, adenotonsillar hypertrophy is present, and the patient has a chronic open-mouth appearance. The parotid salivary flow rate is usually decreased, and eruption of both the deciduous and permanent teeth is delayed in 75% of the cases with irregular sequence of eruption (Brown and Cunningham, 1961). Hypodontia, anodontia, microdontia, posterior crossbite, mandibular-overjet mesioclusion, anterior open bite, crowded teeth, and widely spaced teeth are manifestations of the dental problems these patients have. Occasionally because of the relative macroglossia, reduction glossopexy is indicated; a rhomboid part of the tip of the tongue is removed and a new tip is reconstructed (Cohen and Cohen, 1971). The incidence is 1 in 600 to 1 in 700 births and 1 in 66 of all patients institutionalized for mental deficiency. Associated problems include cardiorespiratory, musculoskeletal, gastrointestinal, and cutaneous manifestations.

Craniofacial dysostosis (Crouzon syndrome) is transmitted as an autosomal dominant trait. This syndrome is secondary to premature craniosynostosis involving the lambdoid, sagittal, and coronal sutures. Manifestations include occasional mental deficiency secondary to increased intracranial pressure, congenital aural atresia with conductive hearing loss, hypertelorism, short upper lip, relative mandibular prognathism with oligodontia, macrodontia, and widely spaced teeth (Fig. 67-12). Maxillary hypoplasia is common, causing a class III malocclusion with a V-shaped dental arch, crowding of the teeth, crossbite, and open bite (Peterson and Pruzansky, 1974).

Acrocephalosyndactyly (Apert's syndrome) (Peterson and Pruzansky, 1974) is secondary to a defect in tissues important in bone development before the fith to sixth week of embryonic life. The syndrome can be secondary to an autosomal dominant transmission; occasionally, sporadic cases associated with increased parental age have been documented. These patients have early obliteration of cranial sutures causing a brachiocephalic or an oxycephalic appearance (see Fig. 67-11) (Park and Powers, 1920). They have syndactyly joint abnormalities and occasionally mental retardation. A high-arched or cleft plata with contriction exists (Fig. 67-12). Approximately 30% of the cases have cleft of the soft palate, hypertelorism, proptosis, and a hypoplastic maxilla, causing a relative prognathism of the mandible and severe class III malocclusion with dental crowding and retarded dental eruption.

Facial hemihypertrophy (Curtius' or Steiner's syndrome) may involve a whole half of the body and is associated with chromosomal abnormalities, endocrine abnormalities, central nervous system lesions, and vascular or lymphatic abnormalities. Asymmetry may be noted at birth but becomes more prominent as the patient grows; it involves the tongue as well as the musculoskeletal structures of the face, such as the lip, palate, maxilla, and mandible. Macroglossia may be unilateral or diffuse, and the lower lip and buccal mucosa may have polypoid excrescences resembling lipomas.

Hemifacial atrophy (Romberg's syndrome) occurs sporadically. Various theories on its cause, especially trauma, have been proposed. The involvement is usually restricted to facial structures. Often it is not noted at birth, but it becomes apparent by 1 year of life.

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Development is retarded, with atrophy in the cartilaginous and musculoskeletal structures causing facial distortion, unilateral enophthalmos, and hemiatrophy of the tongue and half of the upper lip. Maxillary teeth on the involved side are exposed. The body and ramus of the mandible are shorter on the involved side, with delay in development of the angle causing malocclusion.

Type I mucopolysaccharidosis (Hurler syndrome or gargoylism) is transmitted as an autosomal recessive trait with an incidence of 1 in 100.000. The characteristic facial appearance consists of a large head, flattened small nose, prominent forehead, thick earlobes, and thick lips with a flattened philtrum and long upper lip. These characteristics start appearing by the third to sixth month and are apparent by 3 years of age. The mouth is usually held open. Adenoid hypertrophy is very common, causing adenoid facies with tongue protrusion. Lip and tongue enlargement is progressive. The mandible is short and broad with short narrow rami, and the temporomandibular joint motion may be limited because of condylar abnormalities. The teeth are widely spaced, and the local areas of bone destruction in the mandible are referred to as dentigerous cyst. Hyperplastic gingivitis and alveolar ridges may exist.

Oteogenesis imperfecta (van der Hoeve's syndrom or de Kleyn's syndrome) appears as one of two types: congenital or tarda. The latter is more common and secondary to autosomal dominant inheritance. It is secondary to failure of collagen maturation. Bergstrom (1977) has described manifestations of this syndrome, including condylar deformities, prognathic mandible, hypoplastic hemimandible, frequent mandibular dislocation, depressed zygoma, and dentinogenesis imperfecta (in which, because of dentin impairment, the tooth crowns are smaller than normal and the color of the teeth darkens with age). Of these patients 50% to 60% have otosclerosis.

Congenital facial diplegia (Mobius' syndrome) is sporadic. Patients have congenital bilateral cranial nerve palsies (cranial nerve VI and VII). In addition to limb anomalies, chest wall defects and mental retardation occur. These patients also have small mouth apertures with drooping angles allowing drooling of saliva, with unilateral tongue hypoplasia and atrophy with fasciculation or a paralysis that causes difficulty with speech and feeding. A hypoplastic mandible also exists.

Oculoauriculovertebral dysplasia (Goldenhar's syndrome) occurs sporadically, with some increased familial incidence. Vertebral anomalies exist along the abnormal structures of the first and second branchial arches. Mandibular condylal and ramus abnormalities, lingual hypoplasia, macrostomia, salivary gland fistulas, parotid agenesis, and cleft lip or palate may appear. The palatal and tongue muscles may be unilaterally hypoplastic or paralyzed.

Chondrodermal dysplasia (Ellis-van Creveld syndrome) is transmitted as an autosomal recessive trait (Biggerstaff and Mazaheri, 1968). A constant finding in all cases is fusion of the middle portion of the upper lip to the maxillary gingival margin, so that no mucobuccal fold or sulcus exists anteriorly. Natal teeth can exist in 25%. Oligodontia is also a constant finding, especially in the mandibular anterior region with notching of the lower alveolar process. Teeth that erupt are usually small, conically crowned, and regularly spaced with hypoplastic enamel.

Asymmetric crying facies syndrome (Fig. 67-14) is transmitted as a multifactorial inheritance with familial occurrence. The condition is unilateral weakness of the lower lip depressors during crying. Associated anomalies may exist in the cardiovascular, genitourinary, and respiratory systems. The weakness develops because of paralysis of the marginal mandibular branch of the facial nerve, or there may be absence of the muscle. This branch supplies the mentalis and quadratus labii inferioris muscle of the lower lip with a 5:1 predilection for the left side. The body defects are usually on the same side as the paralysis. This defect manifests only during crying; it does not interfere with smiling and sucking and does not cause drooling. The lips at rest are usually symmetric. Weakness usually persists and occasionally may diminish (Pape and Pickering, 1972).

Hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease) is an autosomal dominant condition (Rice and Fischer, 1962). The lips and the tongue (dorsum and tip) are sites of telangiectasia in 60% of the cases. The palate, gingiva, buccal mucosa, and mucocutaneous junction are involved in 20%. Of these cases 20% have mouth bleeding; patients with gingival lesions must be careful brushing their teeth because of easy bleeding. Associated problems include epistaxis, which may need septal dermoplasty, and AV malformations in the lung.

Melkersson-Rosenthal syndrome has a familial occurrence and is more common in women (Kettel, 1947). It appears as episodic recurrent facial paralysis, facial edema, and fissured tongue with atrophy of the papillae. Lip swelling with a reddish brown appearance occurs. Recurring edema results in permanent lip enlargement. Defects in taste exist along the anterior two thirds of the tongue (cranial nerve VII). Simultaneously with skin swelling, oral mucosa is affected in a similar way, resulting in a red color with furrows. Swelling of the palate and gingiva can occur.

Multiple nevoid basal cell carcinoma syndrome is transmitted as an autosomal dominant trait (Gorlin and Sedano, 1971). Jaw cysts (odontogenic keratocysts) occur in 65% to 75% of patients. The syndrome is twice as common in the mandible as in the maxilla, especially in the area of the third molars and canines. Occasionally the cysts can be large enough to cause pathologic fractures. Following cyst removal, recurrence is common (40%).

Sturge-Weber syndrome and encephalofacial angiomatosis are sporadic cases of embryologic anomalies with secondary consequences. Involvement of the lips and buccal mucosa with angiomatosis occurs. The tongue involvement causes hemihypertrophy. A gingival vascular hyperplasia can reach a very large size. Ipsilateral premature or delayed teeth eruption can occur (Gyarmati, 1960).

Table 67-1 lists additional syndromes.