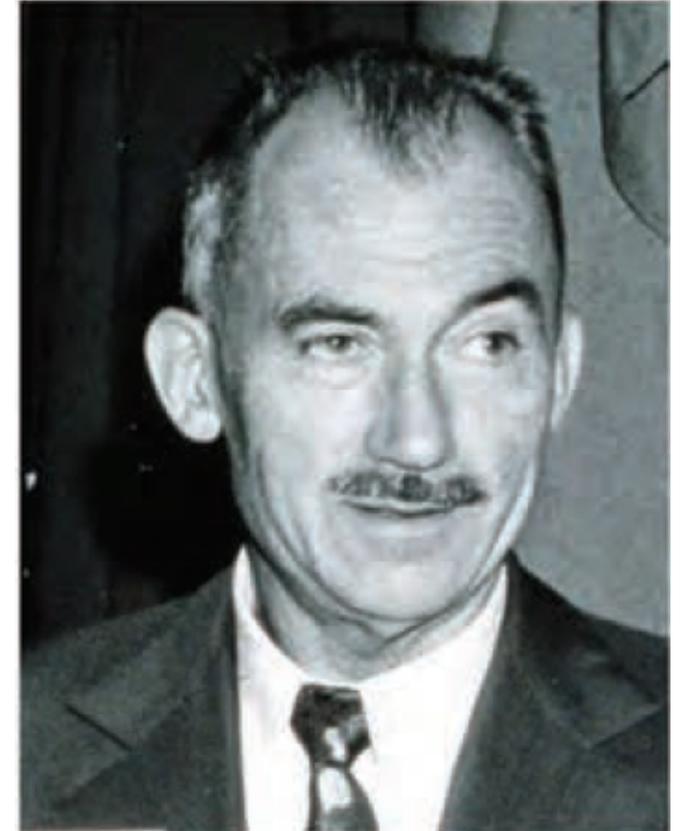


Le Schisi Orofacciali

Roberto Rizzo CMF UNITS

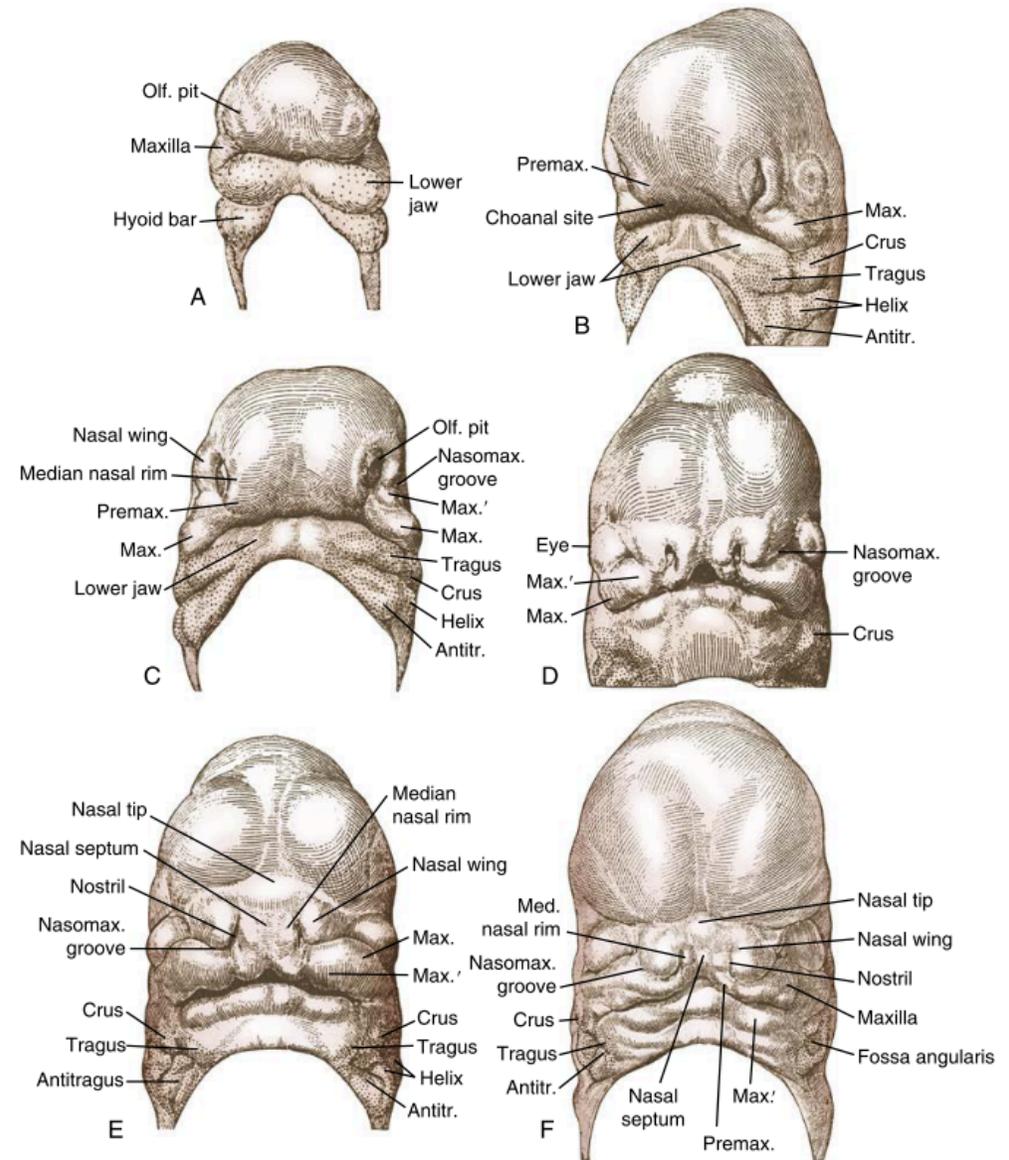
Schisi Orofacciali

- Difetti della complessa morfogenesi facciale
- Le zone di difetto corrispondono a regioni morfologicamente distinte o a centri di crescita embrionale della faccia che sono andate incontro a mancata **fusione** e/o **crescita**
- Classificazione dei difetti di **Paul Tessier**



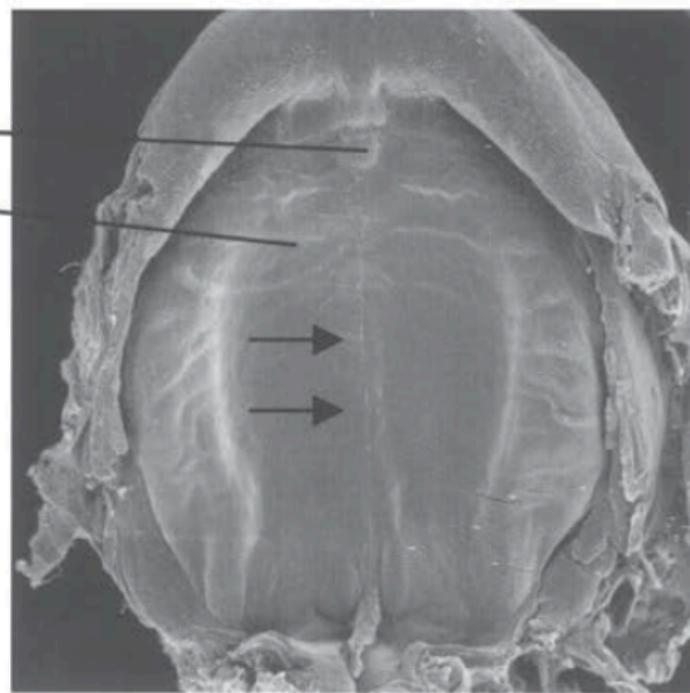
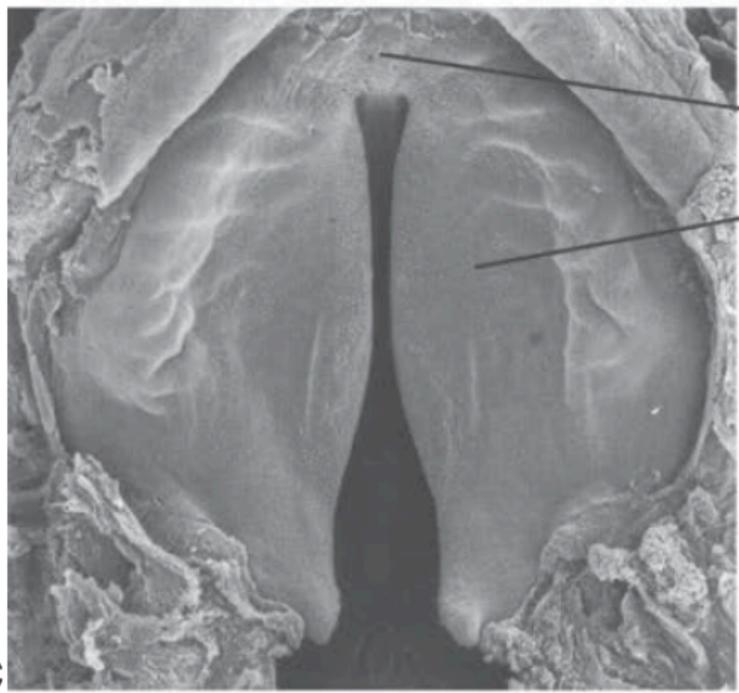
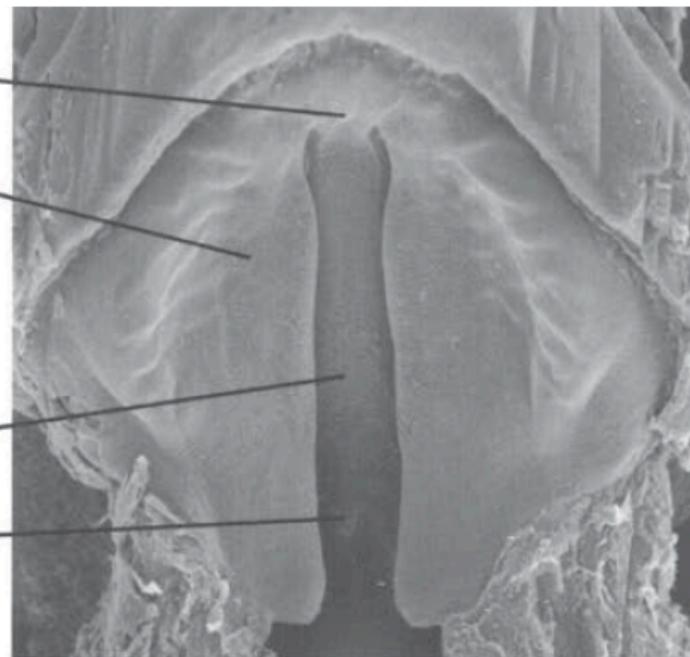
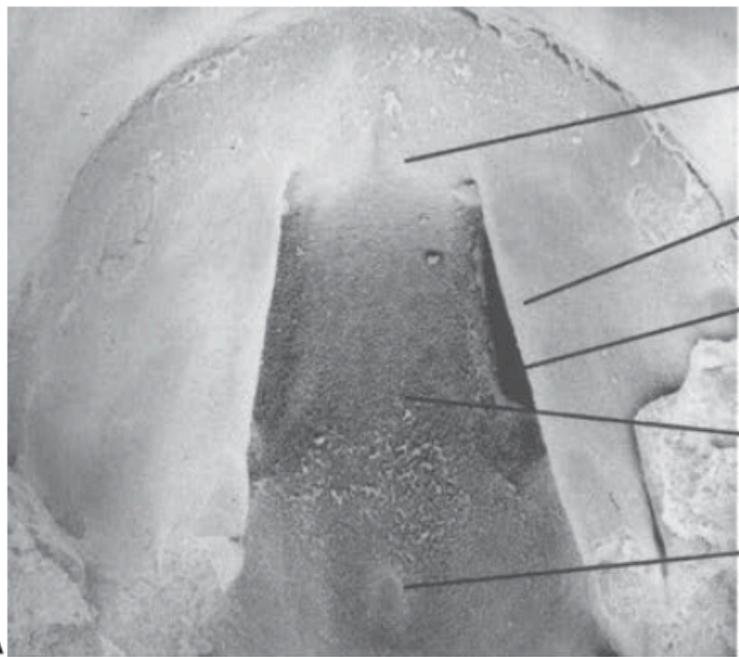
Cenni di Embriogenesi

- **Prominenza Frontonasale:** fronte, naso, filtro del labbro superiore, regione incisale, palato primario.
- **Primi archi branchiali:** processi mascellari, mandibola, antielice, trago.
- **Secondi archi branchiali:** elice, osso ioide.



• **Figure 27-1** Drawings made by J.F. Didusch of human Carnegie stages (Streeter's horizon) 14 (32 days; A), 16 (37 to 40 days; B and C), 17 (41 to 43 days; D and E), and 18 (44 to 46 days; F) from reconstructions of the face. *ANTITR.*, Antitragus; *HYOID BAR*, pharyngeal arch II; *MAX.*, maxillary growth center; *MAX'*, supplementary maxillary growth center; *MED. NASAL RIM*, medial nasal prominence (MNP); *NASOMAX. GROOVE*, nasomaxillary groove; *NASAL WING*, lateral nasal prominence (LNP); *OLF. PIT*, olfactory pit; *PREMAX*, premaxillary center (globular process of His). (Reprinted from Streeter GL: Developmental horizons in human embryos: age groups XV, XVI, XVII and XVIII, being the third issue of a survey of the Carnegie Collection, *Contrib Embryol Carnegie Inst Wash* 32:133-203, 1948.)

- La chiusura bilaterale del labbro superiore determinata dall'unione della premaxilla con la maxilla e con il processo frontonasale avviene nella **SESTA SETTIMANA** di gestazione
- Dalla maxilla si approfondiscono verso l'interno i due palati secondari
- La fusione tra i palati secondari ed il palato primario avviene alla **NONA SETTIMANA**



PP
SP
POSTERIOR
NASAL
CHOANA
NS
RP

PP
SP

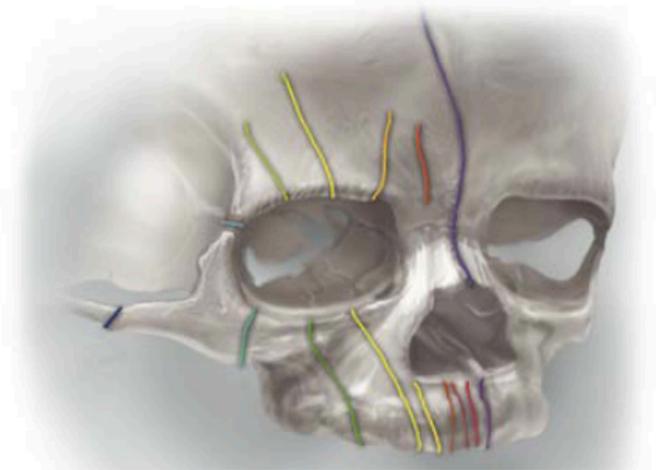
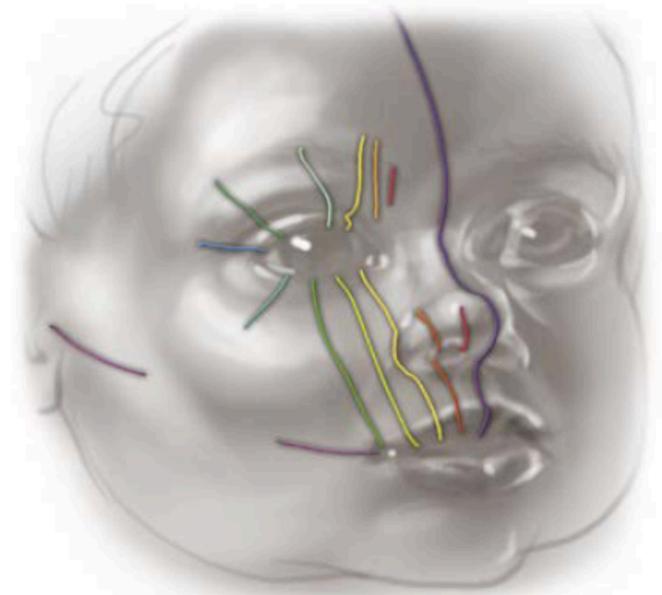
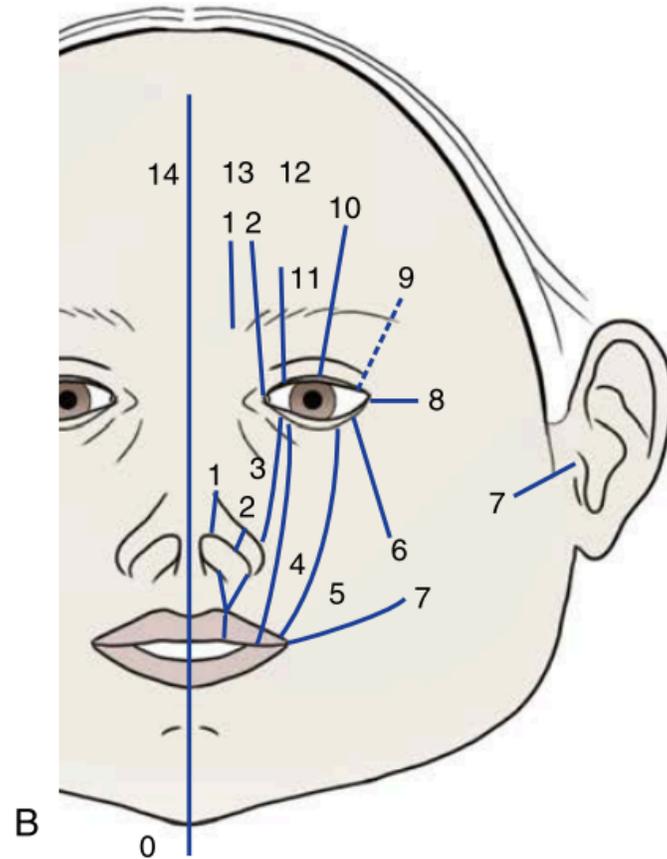
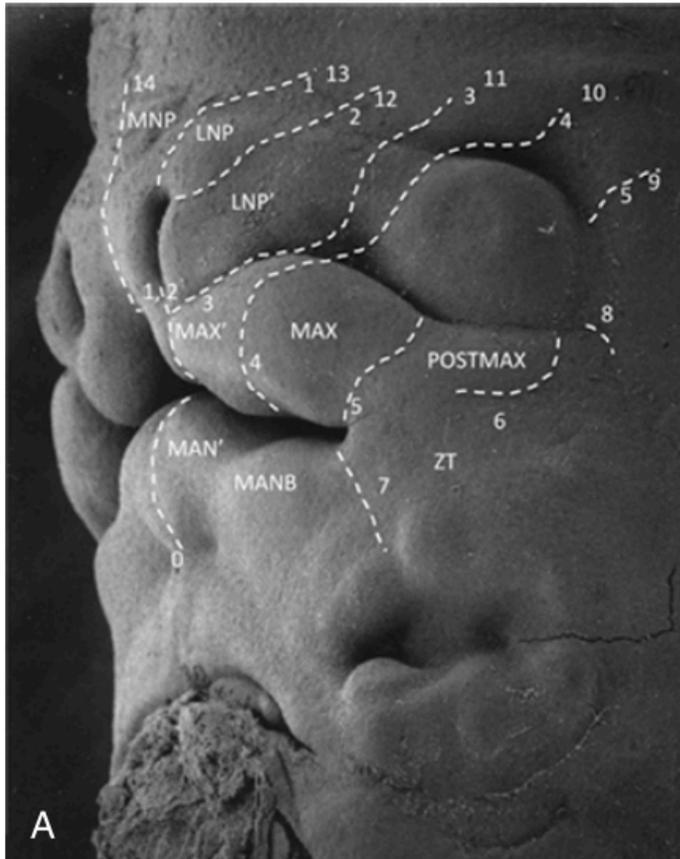
A

B

C

D

Siti delle Schisi Facciali: schema orbitocentrico di Tessier



• **Figure 27-7** Furrows between facial growth centers, most of which remain visible in the 7 week old human embryo shown in (A) are correlated to the position of Tessier's numbered facial clefts (B). (Reprinted from Tessier P: Anatomical classification facial, cranio-facial and latero-facial clefts, *J Maxillofac Surg* 4:69-92, 1976.)

• **Figure 28-2** Complex facial clefts can be classified according to Tessier's original orbitocentric system of numbering. Clefts may involve all tissue planes, including skin, mucosa, bone, teeth, muscle, brain, peripheral nerve, and other specialized tissues.

Terminologia

- **Labioschisi:** difetto di cute, muscolo e mucosa, può estendersi dal naso al vermiglio, può essere completo o incompleto
- **Microschisi:** un solco lungo il labbro superiore con o senza interruzione del muscolo



Incomplete cleft lip.



Complete cleft lip.



Microform cleft lip.



Terminologia

- **Palatoschisi:** difetto di muscolatura del palato molle, con o senza difetto della mucosa, può coinvolgere anche il palato duro ed il processo alveolare.
- **Barra di Simonart:** mucosa nasale unita alla cute in corrispondenza di una schisi



Cleft palate.



Simonart's band.

Terminologia

- **Schisi Sottomucosa:** difetto della sola muscolatura del palato molle senza difetto della mucosa.
- **Incompetenza Velofaringea:** impossibilità per il palato molle difettoso di chiudere il nasofaringe separandolo dall'orofaringe; determina voce nasale
- **Fistola Palatale:** comunicazione tra cavità nasale e orale



Palatal fistula.

Terminologia

- **Fistola oronasale:** comunicazione tra pavimento nasale e vestibolo orale.
- **Schisi Alveolare:** difetto del processo alveolare superiore tra incisivo laterale e canino



Alveolar cleft with oronasal fistula.

Problematiche delle schisi

- **Alimentazione** difficoltosa e facilità di rigurgito
- **Deformità** associate (cardiache, respiratorie)
- Parola e sviluppo del linguaggio (ipernasalità, pronuncia)
- **Otiti** ricorrenti per malposizione delle tube
- **Malocclusione, disgnazia**, ipoplasia mascellare con sviluppo in classe III
- **Difetto estetico** del labbro, del naso, del volto

Etiologia delle Schisi Orofacciali

- **Fattori genetici** che si aggiungono a **fattori ambientali**
- Possono essere associate ad altre sindromi genetiche
- Vengono definite nonsindromiche se non sono associate a ritardo mentale

Table 33.1 Genes associated with orofacial clefting

Candidate gene	Gene map locus	Name of gene	Type of cleft associated
TGF- α	2p13	Transforming growth factor alpha	CL/Cp, CPO
TGF- β	19q13.1–q13.3	Transforming growth factor beta	CL/CP
MSX-1	4p16.1	Homeobox gene (HOX7)	CL/CP
RAR- α	17q12	Retinoic acid receptor alpha	CL/CP
DLX-2	2q32	Distal-less homeobox 2	CPO
BCL-3	19q13.1	B-cell leukemia/lymphoma 3	CL/CP
	2q32	Unknown gene but different than DLX2; possible candidate gene include FN1, IHH, IGFBP2 and IGFBP5	CPO
	4q25–q31.1	Unknown gene	CL/CP
	6q23	Unknown gene	CL/CP
	17p11.1–p11.2	Unknown gene that may increase cleft palate susceptibility or work synergistically to increase susceptibility in Van der Woude syndrome that maps to 1q32–q41	CPO

CL/CP, cleft lip with or without cleft palate; CPO, cleft palate only.

Etiologia delle Schisi Orofacciali: Fattori Ambientali

1. Smoking
2. Alcohol intake (ethyl alcohol)
3. Drugs such as phenytoin, diphenyl hydantoin, methotrexate, aminopterin, retinoids and sodium valproate
4. Pesticides such as dioxin
5. Hyperthermia during pregnancy

Etiologia: sindromi associate

Syndromes associated with cleft lip and palate

1. Chromosomal
 - a. Trisomy 13
 - b. Trisomy 18
 - c. Velocardiofacial syndrome (22q11 deletion)
2. Nonmendelian
 - a. Pierre Robin sequence
 - b. Goldenhar syndrome (Fig. 33.15)
3. Mendelian disorders
 - a. Ectrodactyly-ectoderm 11
 - b. Dysplasia-clefting syndrome (AD)
 - c. Gorlin syndrome (AD)
 - d. Oto-palato-digital syndrome (XL)
 - e. Oral-facial-digital syndrome (XL) (Fig. 33.16)
 - f. Smith-Lemli-Opitz syndrome (AR)
 - g. Stickler syndrome (AD)
 - h. Treacher Collins syndrome (AD) (Fig. 33.17)
 - i. Van der Woude syndrome (AD) (Fig. 33.18)
4. Unknown
 - a. De Lange syndrome
 - b. Kabuki syndrome
5. Teratogenic
 - a. Foetal alcohol syndrome
 - b. Foetal phenytoin syndrome
 - c. Foetal valproate syndrome

AD, autosomal dominant; AR, autosomal recessive; XL, X-linked inheritance.

Prevalenza

- 1 ogni 700 nati
- Più colpito il sesso **maschile**
- Più colpito il lato **sinistro**
- **Unilaterale** più frequente del bilaterale
- Solo palato 1 ogni 2000 nati
- Difetto **evidenziabile ecograficamente** alla 16° settimana di gestazione

Ereditabilità

- Se il primo figlio ha la schisi, il secondo ha 30-40x il rischio di svilupparla rispetto alla popolazione generale

Nutrizione

- Bambini con solo labioschisi monolaterale possono alimentarsi al seno oppure dal biberon con tettarella modificata.

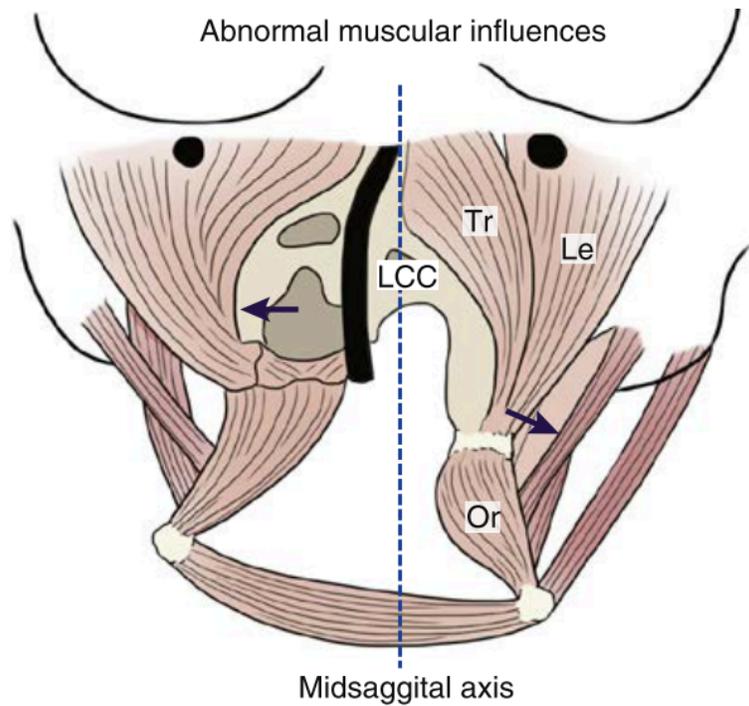
Anatomia Patologica schisi monolaterale

- Naso ruotato dal lato sano
- Columella ruotata nella direzione opposta
- Pavimento nasale assente
- Setto deviato verso il lato opposto
- Muscolo orbicolare interrotto

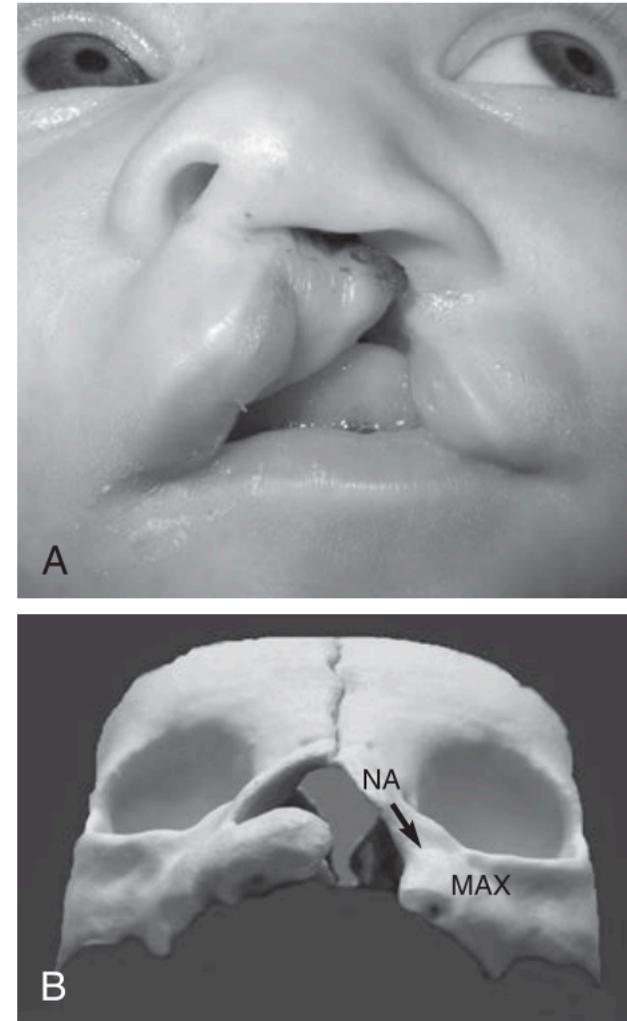


Fig. 33.19 A Incomplete UCLP. B Complete UCLP.

Anatomia Patologica schisi monolaterale



• **Figure 29-25** Schematic of muscles in a cleft model that demonstrates the abnormal position and influence of the disrupted musculoaponeurosis. *Arrows* indicate the muscle pull on the nasal septum and ala with consequent distortion. *Tr*, Transversalis; *Le*, levator; *Or*, orbicularis; *LLC*, lower lateral cartilage. *Dotted line*, Midsagittal axis.



• **Figure 29-26** **A**, Newborn demonstrating distortion of facial structures by the underlying disrupted muscles. **B**, Model of cleft in newborn demonstrating hypoplasia and distortion of bony skeleton in direction of *arrow*. *NA*, Nasal bone; *MAX*, maxilla.

Anatomia Patologica schisi bilaterale

- Il **segmento prenasale** non è unito al mascellare ed è ruotato anteriormente
- Il **setto** nasale è diritto e la columella assente
- La base dell'**ala nasale** è appiattita
- Pavimento nasale assente bilateralmente
- Muscolo **orbicolare** interrotto ed assente nella porzione anteriore



Fig. 33.20 Bilateral clefts: **A** Incomplete bilateral cleft. **B** Complete bilateral cleft.

Anatomia Patologica schisi palatina

- **Muscolatura** del palato molle interrotta al centro e deviata lateralmente e con inserzioni anomale
- Assenza di mucosa ed osso lungo il rafe palatino

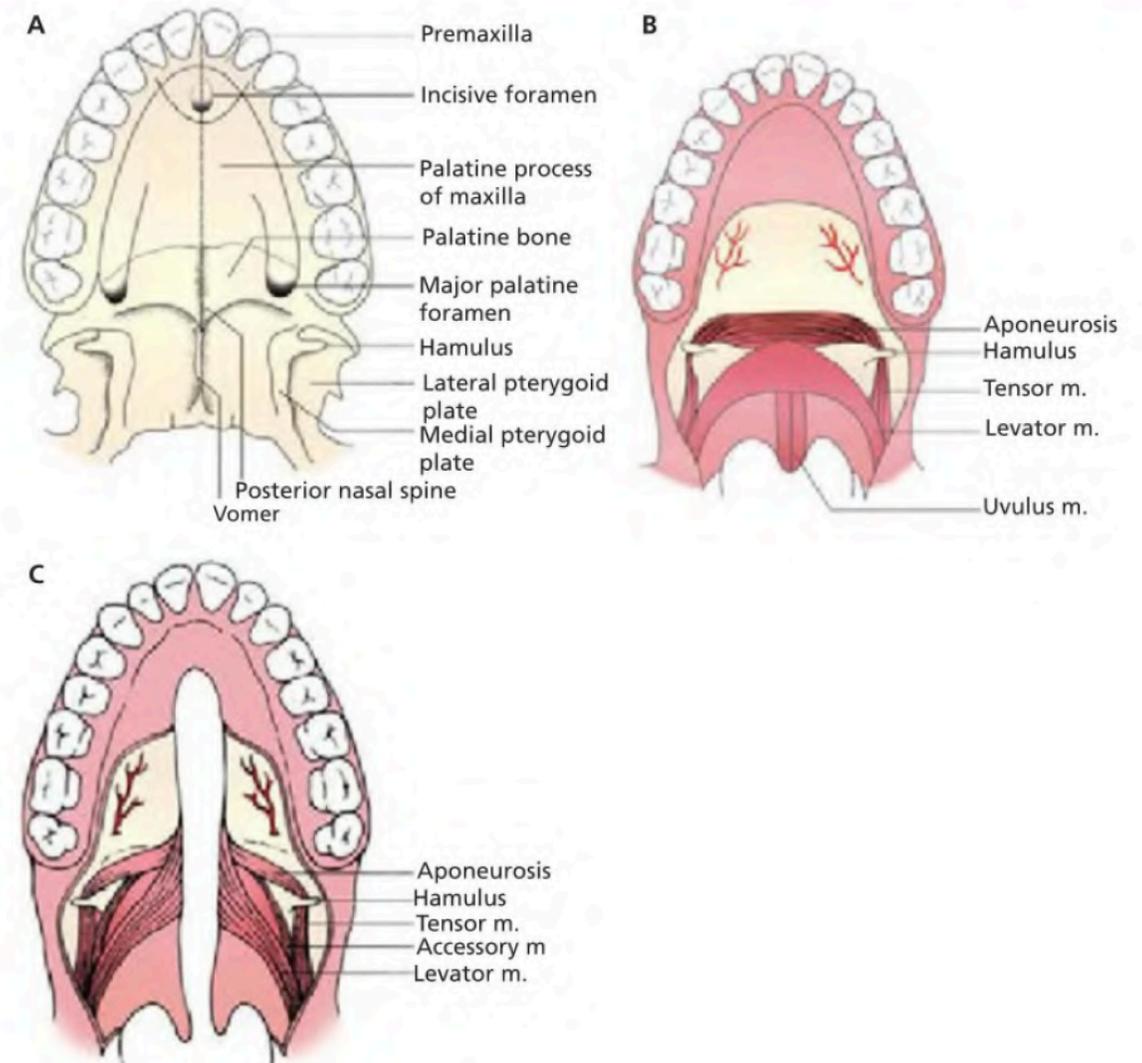
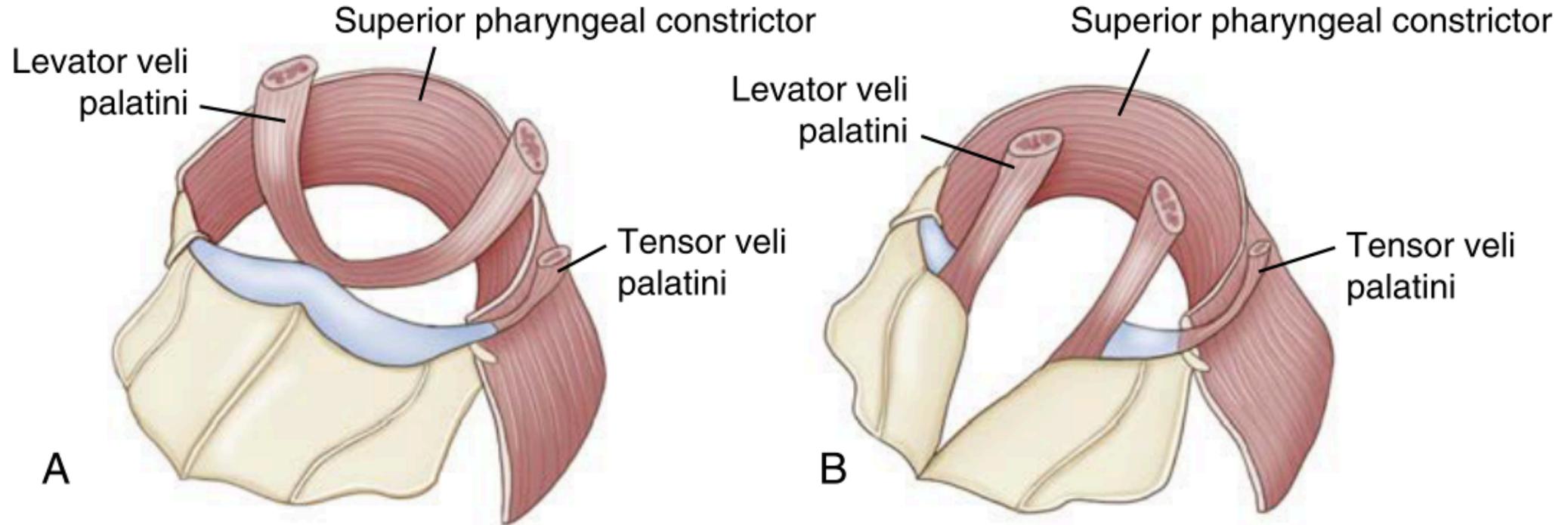


Fig. 33.21 A, B Normal anatomy of the palate. C Abnormal anatomy of palate.

Anatomia Patologica Schisi Palatina



- **Figure 30-1 A**, The key anatomic components of the normal palatal and velopharyngeal anatomy are displayed, including the intact posterior hard palate and vomer, levator and tensor sling, and superior pharyngeal constrictor which forms the posterior portion of the velopharyngeal valve. **B**, The key anatomic components of the abnormal palatal and velopharyngeal anatomy associated with cleft palate are displayed, including the hard palate defect and absent vomer, division of the levator sling, and abnormal insertion of the tensor.

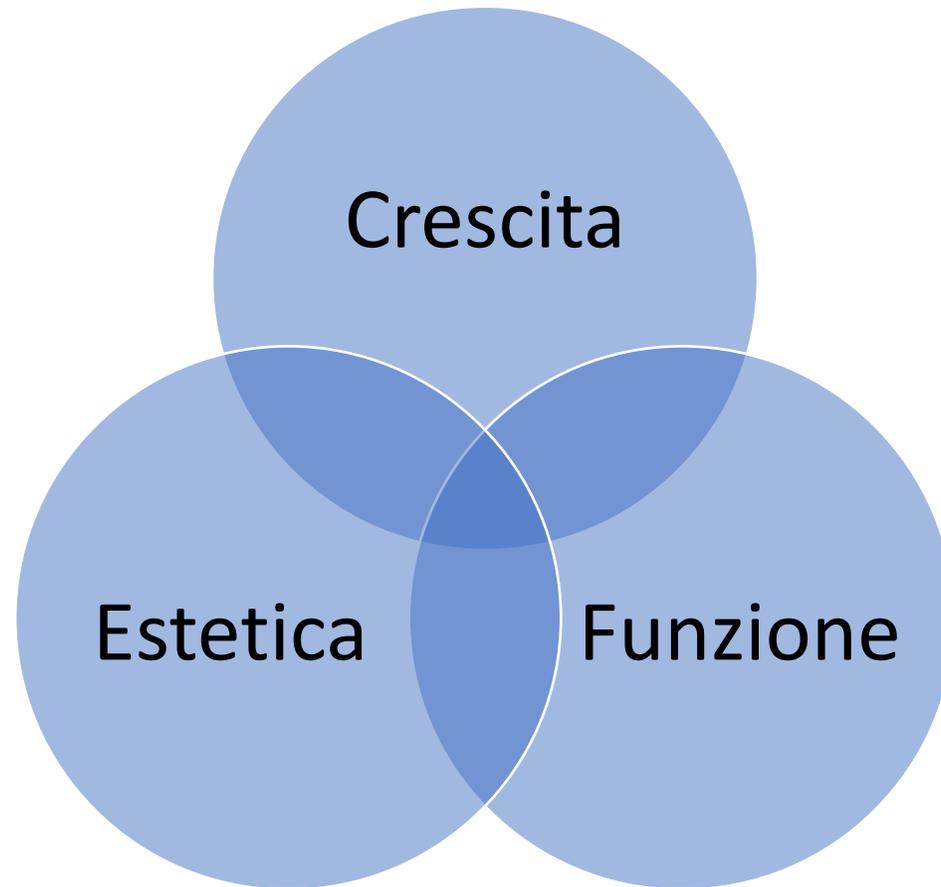
Gestione complessa delle schisi

MANAGEMENT OF PATIENTS WITH CLEFT LIP AND PALATE

SEQUENCE OF PROCEDURES

1. Primary
 - a. Feeding plate/presurgical alveolar moulding
 - b. Primary lip and nose repair
 - c. Primary palatal repair
2. Secondary
 - a. Secondary alveolar bone grafting
 - b. Pharyngoplasty
 - c. Orthodontic treatment
 - d. Orthognathic procedures/distraction osteogenesis
 - e. Rhinoplasty and scar revision of the lip

Timing del rattamento delle schisi:
bilanciamento tra...



Timing

TABLE 28-1 Staged Reconstruction of Cleft Lip and Palate Deformities

Procedure	Time Frame
Cleft lip repair	After 10 weeks
Cleft palate repair	Age 9-18 months
Pharyngeal flap or pharyngoplasty	Age 3-5 years or later based on speech development
Maxillary/alveolar reconstruction with bone grafting	Age 6-9 years based on dental development
Cleft orthognathic surgery	Age 14-16 years in girls, 16-18 years in boys
Cleft rhinoplasty	After age 5 years, but preferably at skeletal maturity; after orthognathic surgery when possible.
Cleft lip revision	Anytime once initial remodeling and scar maturation is complete, but best performed after age 5 years

Labioschisi: timing dopo le 10 settimane

- Per avere il tempo di eseguire una valutazione approfondita
- Per avere reperi anatomici meglio definiti
- Regola del 10 (10 settimane, 10 libbre, 10 mg/dL emoglobina)
- Prima delle 10 settimane le cicatrici risultano più evidenti

Palatoschisi: timing tra 9 e 18 mesi

- Combinazione tra restrizione alla crescita facciale prodotta e sviluppo della parola che richiede un palato integro a 18 mesi.
- In caso di ritardo di sviluppo si può attendere di più
- Intervenire prima si rischia un iposviluppo mascellare

Insufficienza Velofaringea: timing 3-5 anni

- Diagnosticata in genere tra i 3 ed i 5 anni e operabile con velofaringoplastica

Schisi del processo alveolare: timing

- Interessa il 75% chi ha una schisi
- La tempistica di intervento è in relazione allo sviluppo dentario
- PRIMA DELL'ERUZIONE DEL CANINO PERMANENTE (12 aa)
- Innesto di osso autogeno prelevato dalla cresta iliaca, dalla sinfisi mentoniera, dalla tibia o dalle coste.

Correzione ortognatica: timing 14-18 aa

- Severa ipoplasia mascellare
- Tra i 14 ed i 18 anni
- Osteotomia di Le Fort 1 ed avanzamento mascellare con o senza
- Osteotomia mandibolare con retrusione
- Rinoplastica

Schisi: pretrattamento chirurgico

- Taping
- Orthopedic

Taping elastico-compressivo

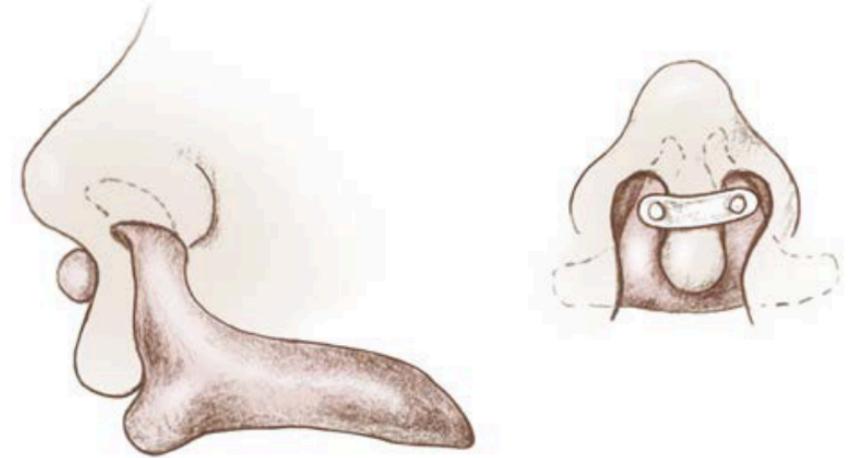
- Per conformare i tessuti molli



Fig. 33.23 **A1, A2** Strapping of bilateral cleft lip using rubber bands to orthopaedically manipulate the prognathic premaxilla to a more favourable position before lip surgery. **B** Strapping also assists in decreasing the width of the cleft and in bringing the lip segments near each other.

Placche in resina acrilica: dibattute

- Latham (100% di ridotta crescita mascellare)
- Grayson (allineatore dei tessuti molli)
- Holtz

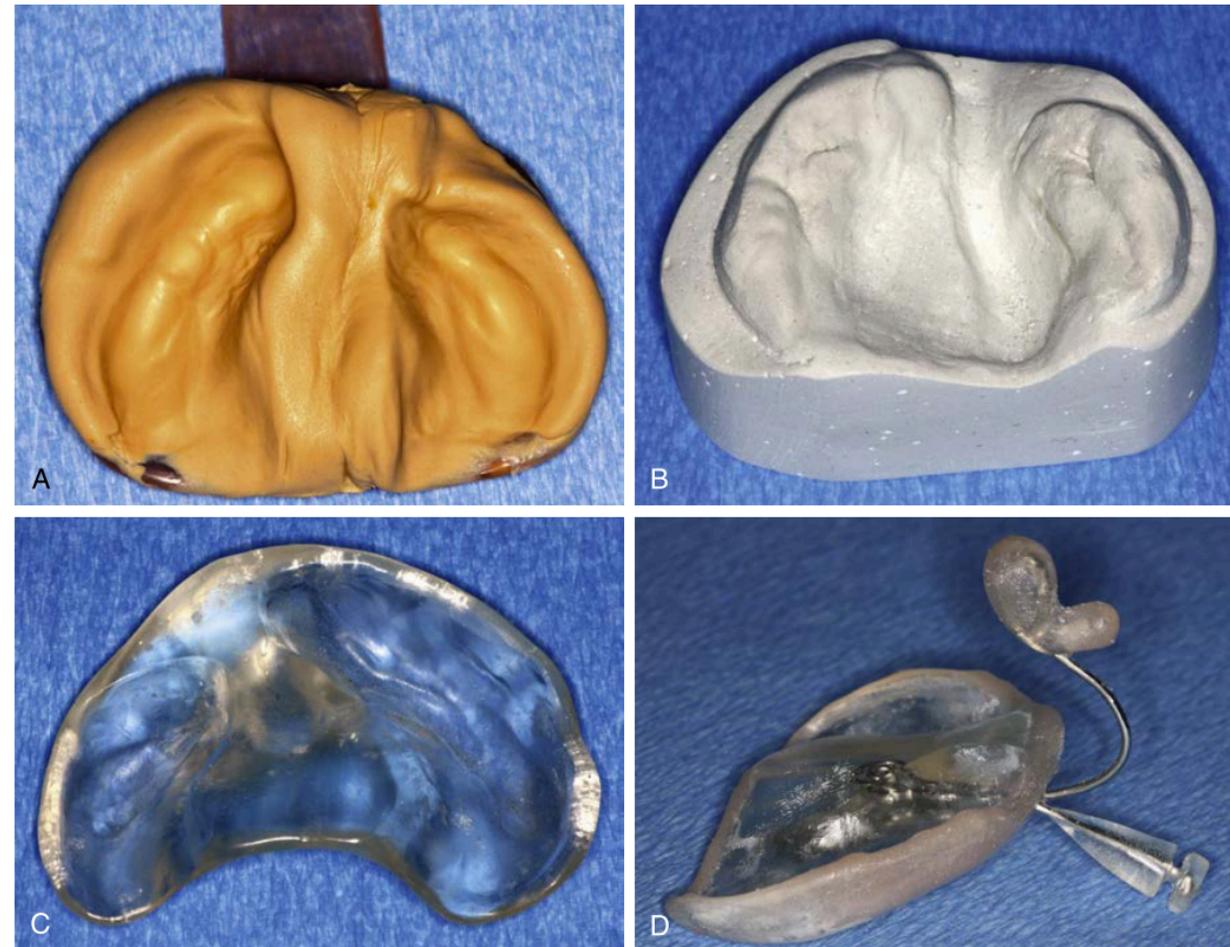


• **Figure 28-3** Frontal and lateral views of the Grayson nasoalveolar molding appliance showing the nasal projections that theoretically help mold the nasal cartilages and maxillary segments into a more appropriate configuration before repair.



Fig. 33.22 Feeding plate or Hotz plate assists in feeding the child without nasal regurgitation can be used as an orthopaedic appliance for moulding the cleft segments to a more favourable, symmetric position before performing lip repair.

Placche in resina acrilica



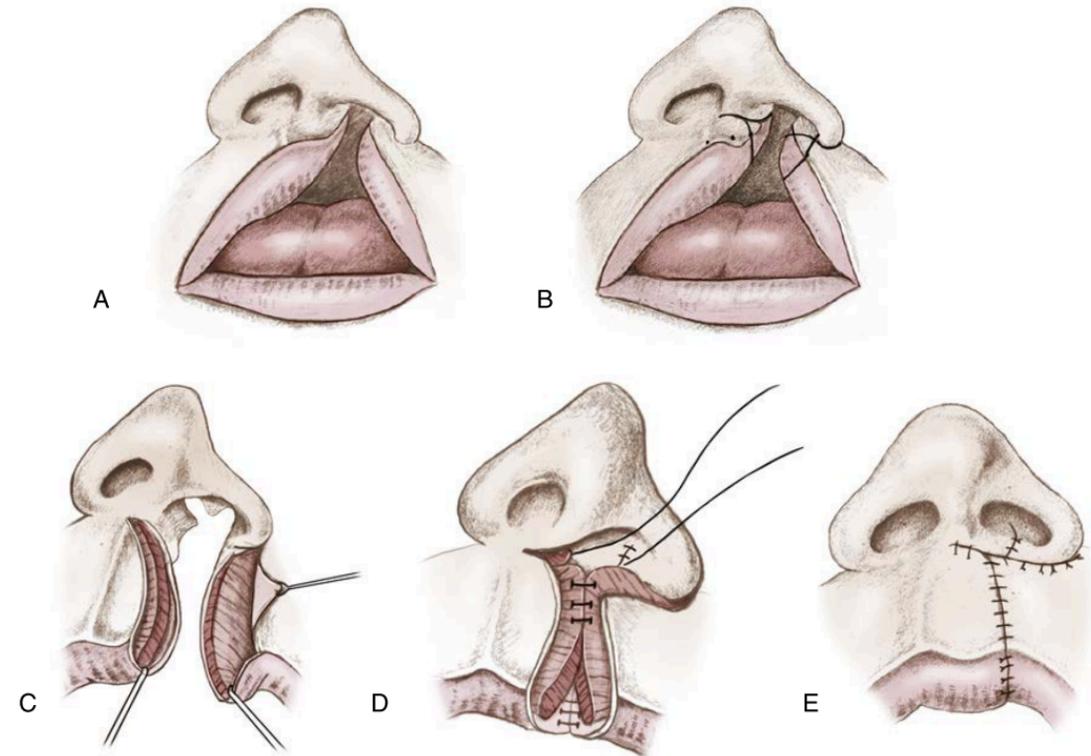
• **Figure 31-1** In the process of making the NAM appliance, a PVS impression is taken of the infant's mouth (A), a stone cast made from this impression (B), and a hard acrylic intraoral plate is fabricated (C). A nasal stent is added to the plate to address the nasal deformity, once the alveolar segments are in close approximation (D).



• **Figure 31-2** The unilateral alveolar, labial and nasal cleft defect severity (A) is addressed through use of the NAM plate (B), accessory transverse lip taping and nasal stenting (C) to normalize anatomy in preparation for the primary surgical repair. NAM improves nasolabial symmetry and esthetics (D).

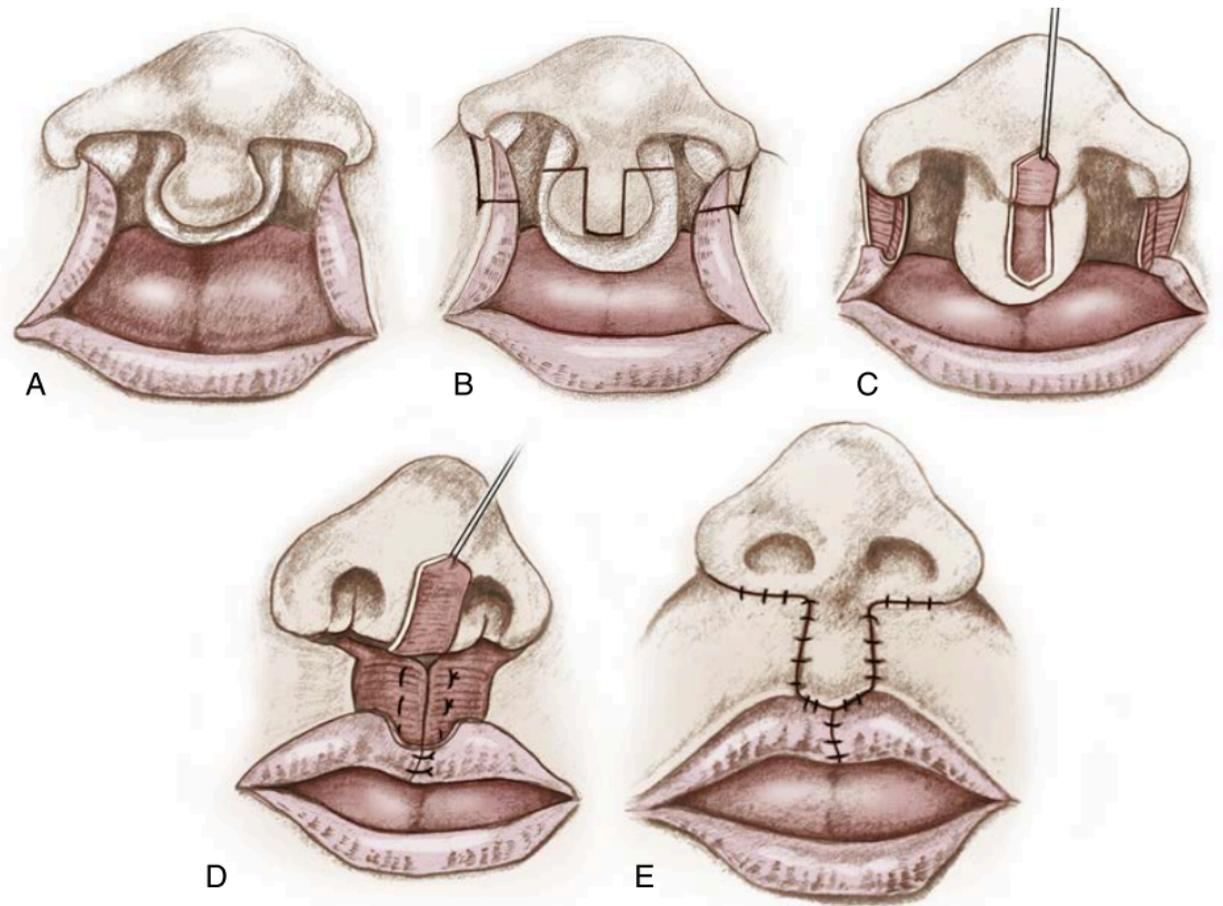
Trattamenti chirurgici labbro: plastica

Creare un triplo strato di cute , muscolo e mucosa
Togliere il tessuto ipoplasico ai margini
Riavvicinare i lembi



• **Figure 28-4** A, Complete unilateral cleft of the lip highlighting the hypoplastic tissue in the cleft site not used in the reconstruction. Note the nasal deformities typical in the unilateral cleft, including displaced lower lateral nasal cartilages, deviated anterior septum, and nasal floor clefting. B, The typical markings for the authors' preferred repair, highlighting the need to excise the hypoplastic tissue and approximate good vermilion and white roll tissue for the repair. C, Once the hypoplastic tissue has been excised, the three layers of tissue are dissected (skin, muscle, and mucosa). Completely freeing the orbicularis oris from its abnormal insertions on the anterior nasal spine area and lateral ala is important. Nasal flaps also are incorporated into the dissection to repair the nasal floor (not shown). D, The orbicularis oris muscle is approximated with multiple interrupted sutures, and the vermilion border/white roll complex is reconstructed. The nasal floor and mucosal flaps are approximated. E, The lateral flap is advanced and the medial segment is rotated downward to create a healing scar line that will resemble the natural philtral column on the opposite side. The incision lines are hidden in natural contours and folds of the nose and lip.

Trattamenti chirurgici labbro: plastica

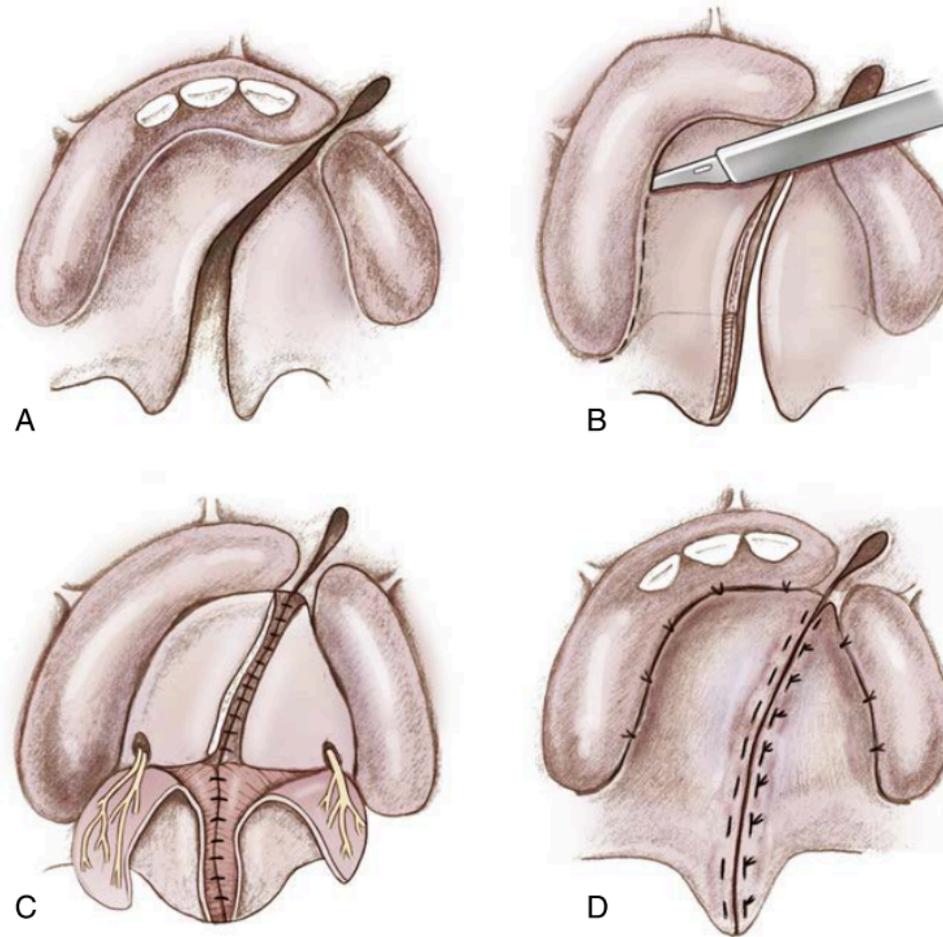


• **Figure 28-7** **A**, The bilateral cleft of the lip and maxilla shown is complete and highlights the nature of hypoplastic tissue along the cleft edges. The importance of the nasal deformity is evident in the shorter columella and disrupted nasal complexes. **B**, Markings of the authors' preferred repair, with an emphasis on excision of hypoplastic tissue and approximating more normal tissue with the advancement flaps. **C**, A new philtrum is created by excising the lateral hypoplastic tissue and elevating the philtrum superiorly. In addition, the lateral advancement flaps are dissected into three distinct layers (skin, muscle, and mucosa). Nasal floor reconstruction also occurs in conjunction with these advancement flaps. **D**, The orbicularis oris musculature is approximated in the midline with multiple interrupted and/or mattress sutures. This is a critical step in the total reconstruction of the functional lip. No musculature is present in the premaxillary segment, which must be brought to the midline from each lateral advancement flap. The nasal floor flaps are sutured at this time as well. The new vermilion border is reconstructed in the midline with good white roll tissue advanced from the lateral flaps. **E**, The final approximation of the skin and mucosal tissues is performed, leaving the healing incision lines in natural contours of the lip and nose.

Trattamenti chirurgici palato: plastica

- Ci si riferisce al **palato secondario**, posteriormente al forame interincisivo, in quanti i difetti del palato primario, al davanti del forame, vengono trattati nell'adolescenza.
- Il trattamento ha per obiettivo 1) la creazione di un **sigillo** tra cavità orale e nasale sia lungo il palato duro che quello molle e 2) la correzione anatomica della **muscolatura** del palato molle che è un punto fondamentale per lo sviluppo della parola.
- 3 strati (mucosa nasale, muscolatura e mucosa orale) per il palato molle, 2 strati mucosi per il palato duro

Palatoplastica



• **Figure 28-12 A**, Unilateral cleft of the primary and secondary palates is shown with the typical involvement from the anterior vestibule to the uvula. **B**, The Bardach palatoplasty technique requires two large, full-thickness mucoperiosteal flaps to be elevated from each palate shelf. The anterior portion (anterior to the incisive foramen) of the cleft is not reconstructed until the mixed dentition stage. **C**, A layered closure is performed in the Bardach palatoplasty by reapproximating the nasal mucosa. The muscle bellies of the levator palatini are elevated off their abnormal insertions on the posterior palate. They are then reapproximated in the midline to create a dynamic functional sling for speech purposes. **D**, Once the nasal mucosa and musculature of the soft palate are approximated, the oral mucosa is closed in the midline. The lateral releasing incisions are quite easily closed primarily because of the length gained from the depth of the palate. In rare cases, in very wide clefts a portion of the lateral incisions may remain open and granulate by secondary intention.

Normale funzione velofarigea

- **Chiusura del rinofaringe** durante la pronuncia di determinati fonemi, durante la suzione, il fischiare, il bere, il deglutire, operata dal palato molle. In particolare dall'azione del muscolo elevatore del velo palatino, del muscolo dell'uvula, del palatofarigeo ed in misura minore del costrittore superiore della faringe.
- Nei difetti, l'aria emessa risuona nel naso e nel nasofaringe.
- Uso di dispositivi protesici temporanei per elevare il palato molle
- Faringoplastica

Difetto alveolare

- Chiusura con innesti di osso autologo ed intervento di gengivoperiostioplastica

GOALS OF SECONDARY ALVEOLAR BONE GRAFTING

- Stabilisation of maxillary arch
- Closure of vestibular and palatal oronasal fistulae
- Provision of bone of sufficient quantity and appropriate quality to allow not only eruption of the permanent lateral incisor and canine teeth but also maintenance of adequate bone about the root of the permanent central incisor adjacent to the cleft
- Provision of support for the soft tissue nasal base and reconstruction of the hypoplastic piriform aperture
- Provision of suitable bony architecture of the premaxilla and anterior face of the maxilla on the cleft side to support accurate nasolabial muscle reconstruction
- Provision of adequate bone stock for ultimate placement of osseointegrated implant
- To provide better periodontal support for teeth bordering the cleft

Trattamento Ortodontico: obiettivo

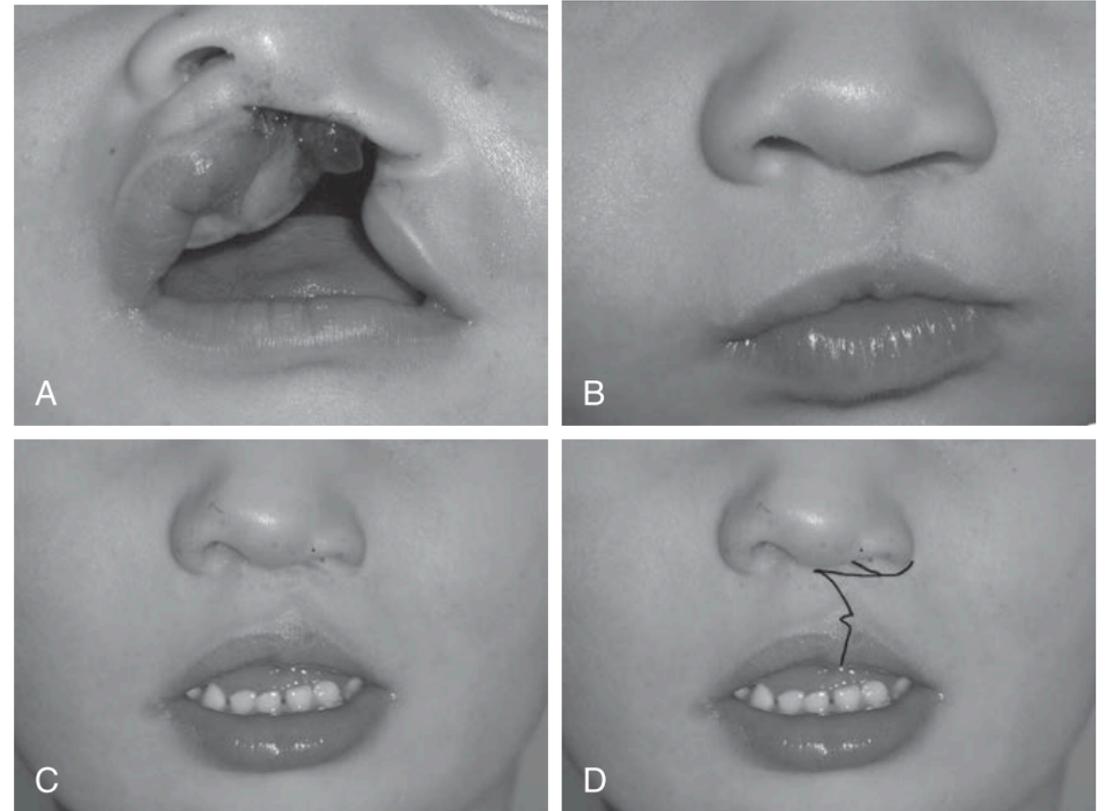
- Consentire un buon allineamento dentario ed una buona funzione
- Espandere l'arcata superiore
- Centrare la linea interincisiva
- Risolvere i crossbite
- Porre le basi per il trattamento chirurgico ortognatico

Chirurgia Ortognatica

- Per risolvere il difetto di crescita (ipoplasia) mascellare ed il rapporto di terza classe che viene a crearsi con la mandibola.
- Avanzamento mascellare di Le Fort I con/senza osteotomia di mandibola

Rinoplastica

- La deformazione nasale **peggiora** con l'età
- La **punta del naso** appare deviata
- Le **narici** sono asimmetriche
- Il **setto** risulta deviato



• **Figure 29-9** Clinical photographs of a patient with a wide, unilateral, complete cleft of the lip and palate. Initial lip repair was performed with a Millard rotation and advancement flap procedure. **A**, A wide cleft defect before closure using the Millard rotation and advancement flap technique. Note the presence of a natal tooth within the cleft. **B**, Six-month postoperative result for the Millard-type cleft repair; the family was diligent in postoperative wound care, massage, and the use of silicone scar gel. **C**, Eighteen-month postoperative results. Note increased nasal asymmetry and some elevation of the vermilion. Child may require minor revision of the nose and vermilion around school age. **D**, Line drawings of the Millard technique used in this patient.

Schisi e Difetti dello sviluppo del linguaggio

- Combinazione di disfunzione velofaringea e compensazione nell'articolazione dei suoni
- Ipernasalità
- Distinguere l'insufficienza velofaringea anatomica dall'incompetenza velofaringea funzionale e dall'uso errato della muscolatura velofaringea

Schisi e Difetti dello sviluppo del linguaggio

**TABLE
32-1**

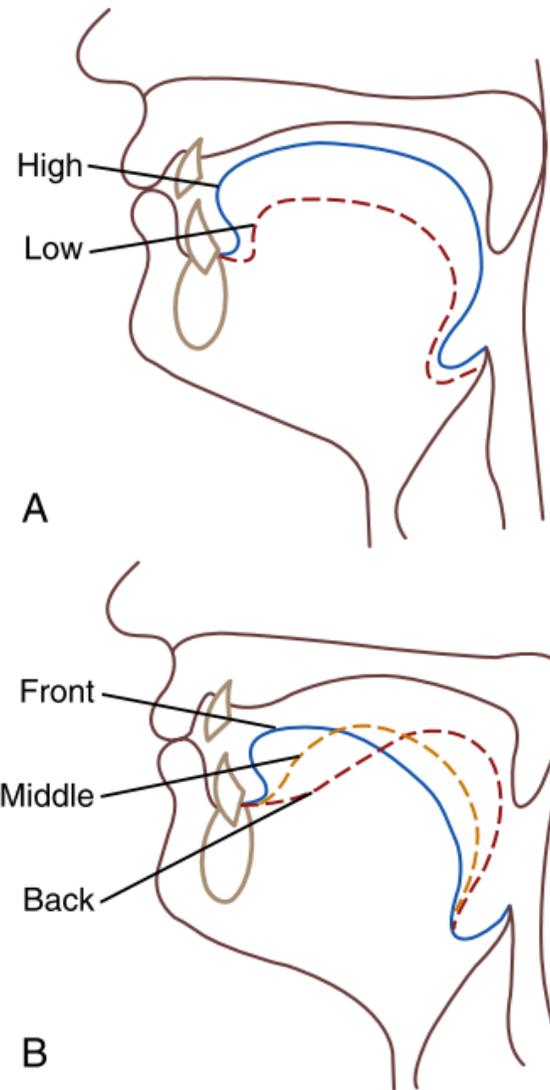
English Consonants Most Likely to Be Affected by Velopharyngeal Dysfunction

	Labial	Labiodental	Interdental	Alveolar	Palatal	Velar
Stops	pb			td		kg
Fricatives		fv	θ ð	sʒ	ʃ ʒ	
Affricates					tʃ dʒ	

**TABLE
32-2**

English Consonants Most Likely to Be Affected by Dental-Alveolar Defects

	Labial	Labiodental	Interdental	Alveolar	Palatal	Velar
Stops				td		
Fricatives		fv	θ ð	sʒ	ʃ ʒ	
Affricates					tʃ dʒ	



• **Figure 32-1** Illustration of normal velopharyngeal closure during vowel production. **A**, Velar and tongue positions for high and low vowels. **B**, Velar and tongue positions for front, middle, and back vowels. (Adapted from Skolnick ML, Cohn ER: *Videofluoroscopic studies of speech in patients with cleft palate*, New York, 1989, Springer-Verlag.)