Università degli Studi di Padova
Dipartimento di Salute della Donna e del Bambino – SDB
U.O.C. Clinica Ginecologica e Ostetrica
Scuola di Specializzazione in Ginecologia e Ostetricia
Direttore Prof. Giovanni Battista Nardelli

LABIOSCHISI E PALATOSCHISI

Dott.ssa Arianna De Lazzari

Caso clinico

L.L. , cinese, 26 aa

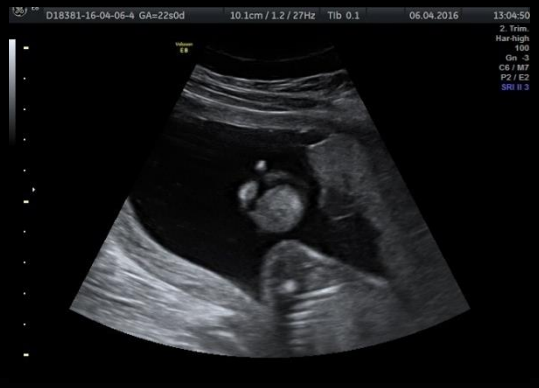
UM 04.11.2015 EPP 10.08.2016

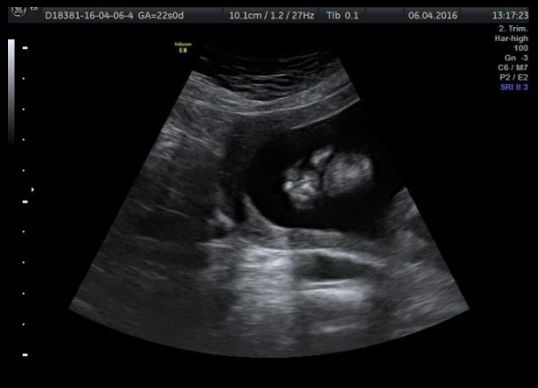
PARA 0010

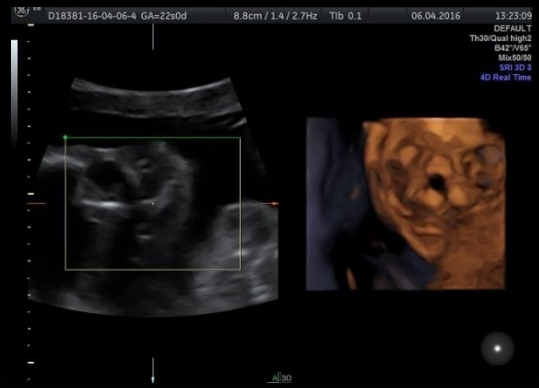
- A. Fam: ndp
- A. Fisio: ndp
- A. Pato: riscontro ipotiroidismo a Febbraio 2016, in tp con Levotiroxina sodica 50 microgrammi/die

Caso clinico

- Eco II trim 31.03.2016(21+1 sg): "*si evidenzia soluzione di continuità di 2,4 mm a carico del labbro superiore con verosimile coinvolgimento della porzione ossea mascellare come da labiopalatoschisi monolaterale sinistra*"
- Eco II trim II livello 06.04.2016 (22 sg): "*si conferma labiopalatoschisi sinistra, che arriva fino alla radice del naso*"







- Diagnosi prenatale invasiva per cariotipo fetale: la coppia rifiuta
- Consulenza chirurgica maxillo-facciale del 07.04.2016: "*…si spiega attentamente il timing dell'intervento che il bambino dovrà affrontare…*"
- Eco di controllo 28.04.2016 (25+1 sg): "*labiopalatoschisi sinistra, interessamento del palato duro per 7,6 x 5,3 mm*"
- OGTT a 26 sg: negativo

Labiopalatoschisi

Labioschisi ± Palatoschisi

Incidenza 1:700 nati vivi

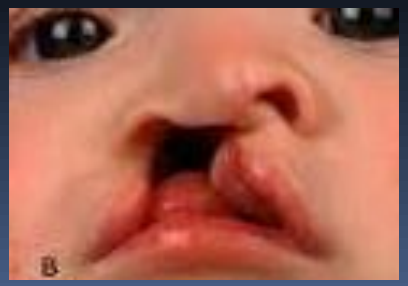
- 1:500 asiatici
- 1:1000 caucasici
- 1:2500 afroamericani

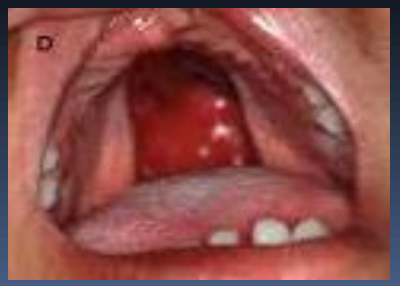
M:F 2:1

- 40% LPS monolaterale (75% sn)
- 29% LS monolaterale
- 26% LPS bilaterale
- 5% LS bilaterale

Palatoschisi Isolata

- Incidenza 1:1500 nati vivi
- M < F



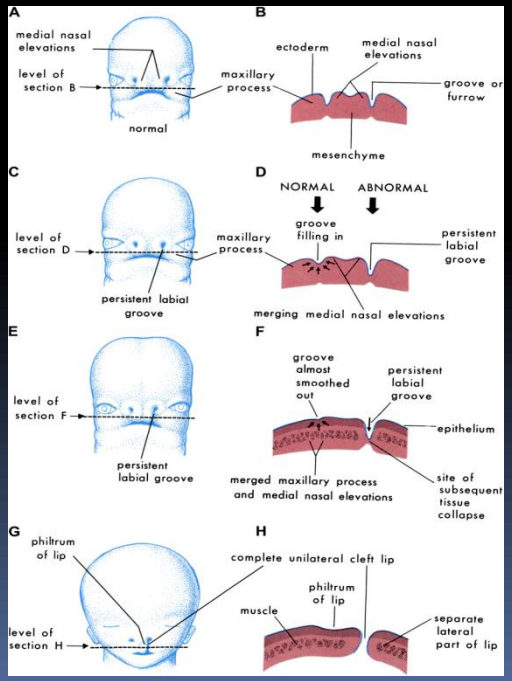


Patogenesi

Le labbra e il palato si sviluppano dal I arco branchiale tra le 7 e 12 sg.

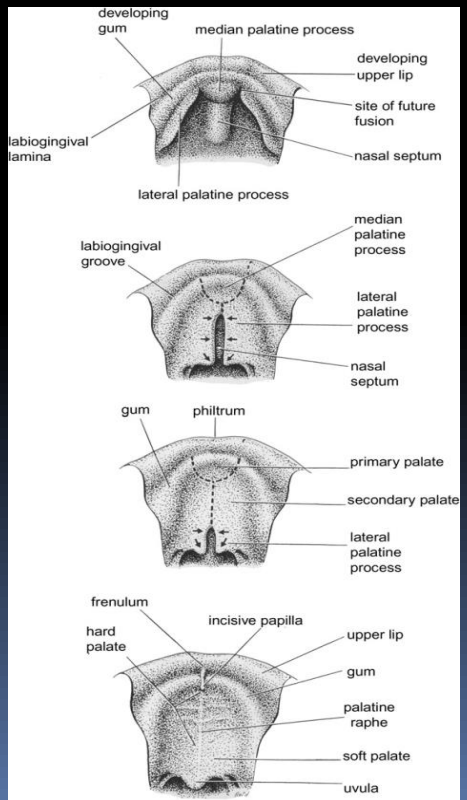
Labbro superiore e filtro:

accostamento dei 2 processi mascellari lungo la linea mediana per fondersi nella parte inferiore del processo nasale mediano

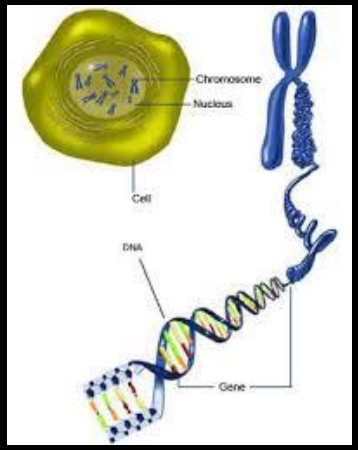


Palato:

fusione mediana dei processi globulari dell'area nasale e dei processi mascellari



Eziologia



- Genetica
  - Difetti di proliferazione
  - Difetti della matrice extracellulare (variante TGF-alfa)
  - Difetti di differenziazione (TGF-beta)
  - Fattori regolatori dell'interferone (IRF-6)
  - Molecole di adesione
- Cromosomica
  - T13
  - T18

Genetic associations with orofacial clefting

	CL±P	CP
Chromosomal	Trisomy 13 or 21	
Single gene	Van der Woude (Chromosome 1, AD)	Treacher Collins Syndrome (Chromosome 5, AD)
	EEC (ectrodactyly, ectodermal hyperplasia and CL±P) Syndrome (Chromosome 3, AD)	Stickler Syndrome (Chromosome 12, AD)
		Velocardiofacial Syndrome (Chromosome 22, AD)
		Opitz G/BBB Syndrome (AD)
Sporadic		Pierre Robin Sequence

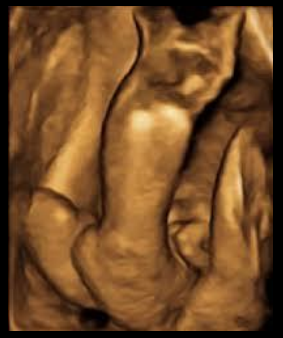
CL±P, cleft lip and palate; CP, cleft palate; AD, autosomal dominant.

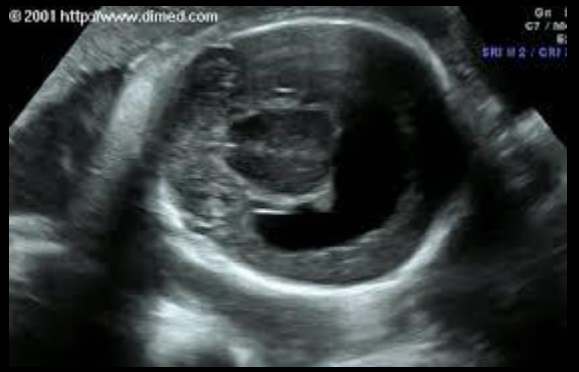
Eziologia

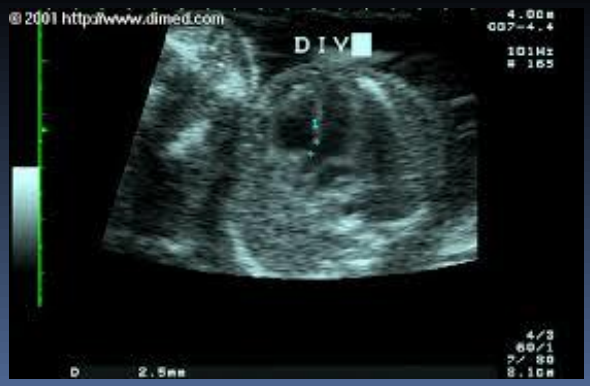
- Ambientale
  - Radiazioni
  - Patologie metaboliche (DM)
  - Teratogeni (Fenitoina, Acido Valproico)
  - Deficit folati (Metotrexate)
  - Fumo
  - Alcol

Anomalie associate

- Cardiopatie congenite
- Scheletriche
- Anomalie cerebrali
- Aneuploidie







Labioschisi isolata	Labiopalatoschisi	Palatoschisi isolata
8%	20%	50%

ELSEVIER SAUNDERS

Otolaryngol Clin N Am
40 (2007) 27–60

OTOLARYNGOLOGIC CLINICS OF NORTH AMERICA

Cleft Lip and Palate

Oneida A. Arosarena, MD

Department of Otolaryngology, Temple University School of Medicine, 3400 North Broad Street, Kresge First Floor, Suite 102, Philadelphia, PA 19140, USA

Table 1
Multiple malformation syndromes associated with cleft lip with or without cleft palate

Genetic disorders	Recognized patterns with unknown genesis	Teratogens[a]
Down syndrome	Amniotic band sequence	Anticonvulsant phenotype
Smith-Lemli-Opitz syndrome	Aicardi syndrome	Fetal alcohol syndrome
Aarskog syndrome	Kabuki make-up syndrome	Maternal diabetes
Coffin-Siris syndrome	Craniofrontonasal dysplasia	Maternal smoking
van der Woude syndrome	Hypertelorism microtia clefting syndrome	Maternal folate deficiency
Waardenburg syndrome	Focal dermal hypoplasia syndrome	—
Ectodermal dysplasia syndromes (Ectrodactyly-ectodermal dysplasia-clefting, Hay-Wells, and Rapp-Hodgkin syndromes)	—	—
Distal arthrogryposis type 2	—	—
Fryns syndrome	—	—
Popliteal pterygium syndrome	—	—
22q deletion syndromes (DiGeorge syndrome, Shprintzen syndrome, and CHARGE association)	—	—
Wolf-Hirschhorn syndrome	—	—
Basal cell nevus syndrome	—	—
Kallman syndrome	—	—
Nail patella syndrome	—	—

[a] Indicates increased risk rather than direct causation [3,6,11–13].

Table 2
Multiple malformation syndromes associated with cleft palate

Genetic disorders	Recognized patterns with unknown genesis	Teratogens[a]
Down syndrome	Pierre-Robin sequence	Anticonvulsant phenotype
Prader-Willi syndrome	Goldenhar syndrome	Fetal alcohol syndrome
Camptomelic dysplasia	Kabuki make-up syndrome	Thalidomide
Stickler syndrome	Mobius sequence	Dioxin
Holoprosencephaly	Klippel-Feil syndrome	Maternal smoking
de Lange syndrome	Silver-Russell syndrome	—
Spondyloepiphyseal dysplasia congenita	Beckwith-Wiedemann syndrome	—
Treacher-Collins syndrome	—	—
Cleft palate–short stature syndrome	—	—
22q deletion syndromes (DiGeorge syndrome, Shprintzen syndrome, and CHARGE association)	—	—
Diastrophic dysplasia	—	—
Orofaciodigital syndrome type I	—	—
Otopalatodigital syndrome type I	—	—
Limb mammary syndrome	—	—
Nager syndrome	—	—
Smith-Lemli-Opitz syndrome	—	—
X-linked cleft palate with ankyloglossia	—	—
Apert syndrome	—	—
Marfan syndrome	—	—
Turner syndrome	—	—
Cleidocranial dysostosis	—	—

[a] Indicates increased risk rather than direct causation [3,6,9,10,56–58].

Ultrasound Obstet Gynecol 2009; 34: 410–415
Published online in Wiley InterScience (www.interscience.wiley.com). DOI: 10.1002/uog.6447

Antenatal detection of cleft lip with or without cleft palate: incidence of associated chromosomal and structural anomalies

J. C. GILLHAM*, S. ANAND† and P. J. BULLEN*

**Fetal Management Unit and †Department of Obstetrics, St Marys Hospital, Manchester, UK*

Table 1 Associated structural and karyotypic abnormalities in unilateral cleft lip with/without cleft palate

		Karyotyping	
Case	*Structural abnormality*	*Time of test*	*Result*
1	Unilateral talipes	Ante	Normal
2	Esophageal atresia	Ante	Normal
3	Unilateral hydronephrosis	Ante	Normal
4	Unilateral dilated renal pelvis	Post	Normal
5	Unilateral talipes	Post	Normal
6	Nephromegaly	Post	Normal
7	ASD	Post	Normal
8	Complex cardiac and echogenic bowel	Ante	Normal
9	Unilateral multicystic dysplastic kidney	Ante	Normal
10	Gastroschisis	Ante	Normal
11	Mild ventriculomegaly	Ante	Normal
12	Anomalous pulmonary veins	Post	Normal
13	VSD, situs inversus	Ante	Trisomy 18 – TOP
14	Agenesis corpus callosum, VSD, horseshoe kidney	Ante	Trisomy 13 – TOP
15	VSD, horseshoe kidney, polydactyly	Ante	Trisomy 13 – TOP
16	Polycystic kidneys	Ante	Trisomy 13 – TOP
17	Choroid plexus cysts, inferior vermian agenesis, univentricular heart	Ante	Trisomy 18 – TOP
18	Polydactyly	Post	Trisomy 13
19	Tracheo-esophageal atresia, hemivertebrae	Ante	TOP; karyotyping refused

Ante, antenatal; ASD, atrial septal defect; Post, postnatal; TOP, termination of pregnancy; VSD, ventricular septal defect.

Table 2 Associated structural and karyotypic abnormalities and genetic syndromes in bilateral cleft lip ± palate

		Karyotyping	
Case	*Structural abnormality*	*Time of test*	*Result*
1	Cord hernia, VM, polydactyly	Ante	Trisomy 18 – TOP
2	IVA, VM, polydactyly, VSD, large echogenic kidneys	Ante	Trisomy 13 – TOP
3	Holoprosencephaly, polydactyly, duplex kidney	Ante	Trisomy 13 – TOP
4	VM, Dandy–Walker malformation, renal cyst	Ante	Normal – TOP
5	Complex cardiac abnormality	Ante	Normal – TOP
6	Holoprosencephaly	Ante	Normal – TOP
7	Bilateral echogenic kidneys	Ante	Normal – TOP
8	Bilateral talipes	Ante	Normal – TOP
9	Abnormal feet, ectrodactyly	Post	Normal
10	ASD, VSD, hypertelorism	Post	Normal
11	Hypertelorism	Post	Normal; craniofrontonasal dysplasia present

Ante, antenatal; ASD, atrial septal defect; IVA, inferior vermian agenesis; Post, postnatal; TOP, termination of pregnancy; VM, ventriculomegaly; VSD, ventricular septal defect.

Results *Over the 6-year period investigated, 570 infants were referred to the FMU and/or CLAP team. Among these, there were 24 terminations of pregnancy, two intrauterine fetal deaths and one early neonatal death identified. Data on 69 of the 543 patients that survived were incomplete. Of 188 cases with unilateral and 34 cases with bilateral cleft lip ± palate there were no karyotypical abnormalities without other structural abnormalities. The incidence of associated structural abnormalities varied with the anatomical type of cleft: that of unilateral cleft lip ± palate was 9.8% (19/194), that of bilateral cleft lip and palate was 25% (11/44) and that of midline cleft lip and palate was 100% (11/11). None of 252 cases with isolated cleft palate was identified antenatally; of these, 5.6% (n = 14) had either karyotypical or associated structural abnormalities and 21.0% (n = 53) had a genetic syndrome as an underlying diagnosis.*

Conclusions *It is essential to tailor the antenatal counseling of patients to the specific scan diagnosis, considering both the anatomical type of cleft and the presence or absence of associated abnormalities. It is inappropriate to offer invasive testing to all patients. The use of three-dimensional ultrasound as an adjunct should be considered in these patients to improve the accuracy of prenatal diagnosis.*

Diagnosi

Vanno ricercate e/o misurate le seguenti strutture:

Estremo cefalico

- Misura del diametro biparietale (BPD) e della circonferenza cranica (CC).
- Misura dell'ampiezza del trigono ventricolare.
- Misura del diametro trasverso del cervelletto.
- Visualizzazione cavo del setto pellucido (CSP).
- Visualizzazione della cisterna magna.

Il CSP diviene visibile a partire dalla 18ª settimana di amenorrea e va incontro a obliterazione verso il termine di gravidanza. E' una struttura la cui non visualizzazione o alterazione è associata a diverse patologie. Si ricorda peraltro che alcune di esse sono di difficile evidenziazione (es. displasia setto-ottica). La cisterna magna va valutata in modo qualitativo rivolta alla ricerca di difetti del tubo neurale e di malformazioni cerebellari.

- Visualizzazione delle orbite.
- Visualizzazione del labbro superiore.

Ultrasound Obstet Gynecol (2010)
Published online in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.8831

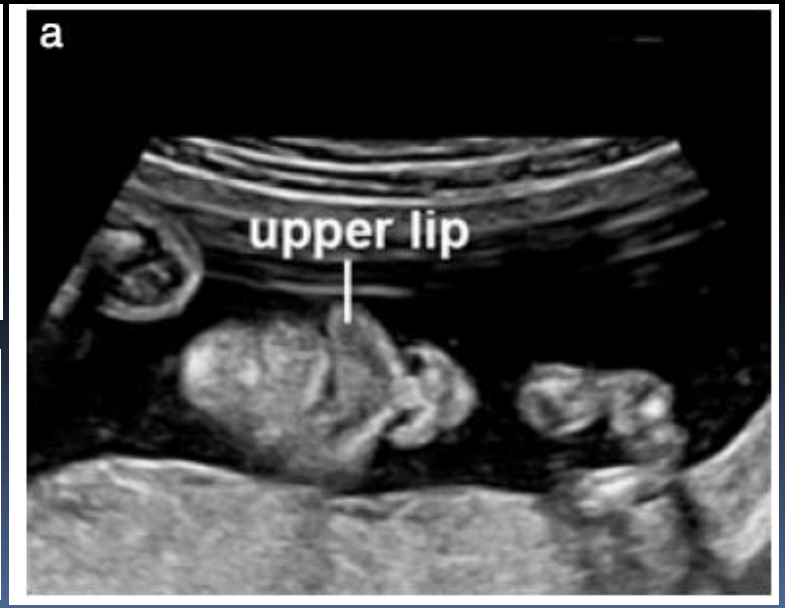
isuog.org

GUIDELINES

Practice guidelines for performance of the routine mid-trimester fetal ultrasound scan

Face

Minimum evaluation of the fetal face should include an attempt to visualize the upper lip for possible cleft lip anomaly[43] (Figure 3a). If technically feasible, other facial features that can be assessed include the median facial profile (Figure 3b), orbits (Figure 3c), nose and nostrils.



Ecografia

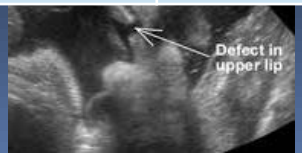
Scansione coronale e obliqua

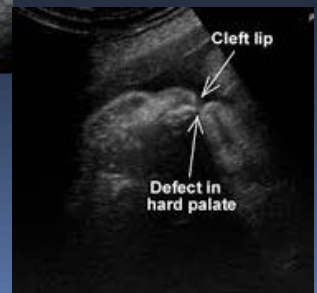
- Valutazione del difetto labiale
- Valutazione dei difetti mediani e delle anomalie del naso
- Bassa attendibilità sui difetti laterali

Scansione trasversale

- Valutazione del difetto osseo
- Difficoltà di rilevazione dei difetti del palato secondario

	N° casi	Sensibilità
EUROCAT 2008-2012	2821	53%
Maarse 2007- 8	38	86%
Ensing 2007-10	76	85%

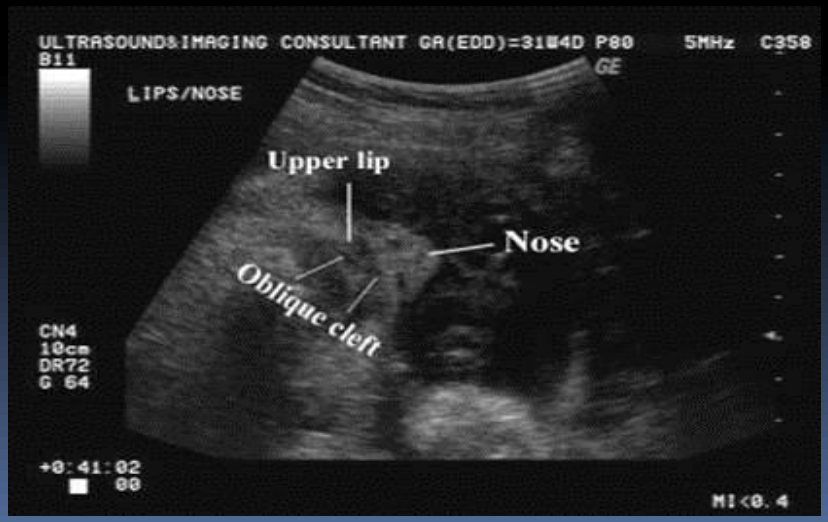


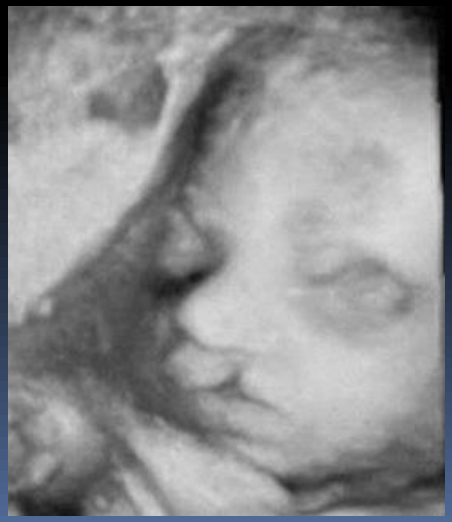


Classificazione di Nyberg

Tipo 1

- Monolaterale
- Coinvolge i tessuti superficiali
- Scansione coronale del labbro superiore
- Non associato a difetti della linea mediana
- Raramente associato ad aneuploidie o ad altre anomalie

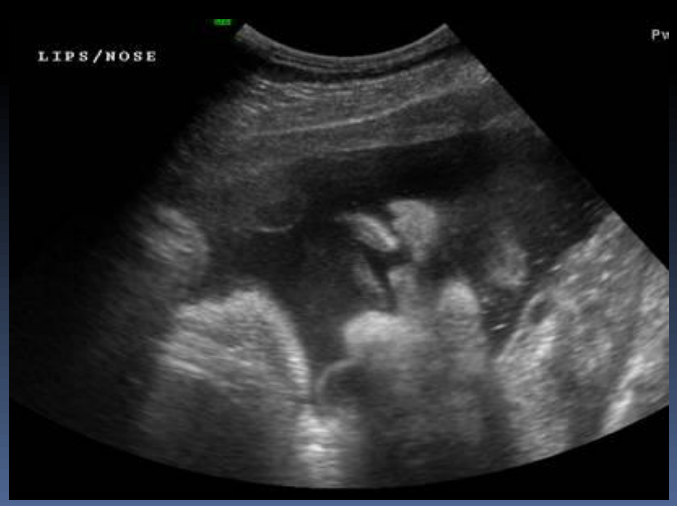


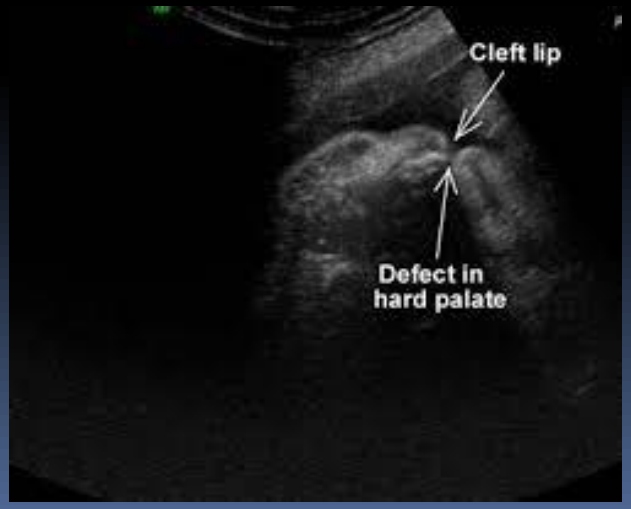


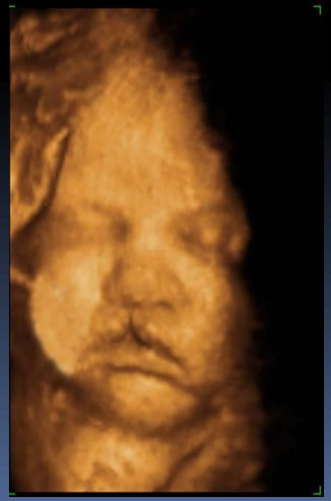
Classificazione

Tipo 2

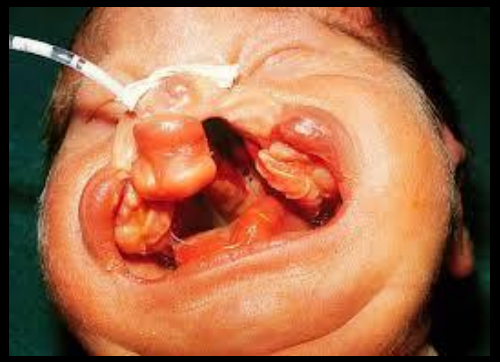
- Monolaterale
- Difetti profondi con distorsione delle strutture ossee
- Scansione coronale e trasversale
- Nel 32% associato ad aneuploidie, nel 48% ad altre anomalie





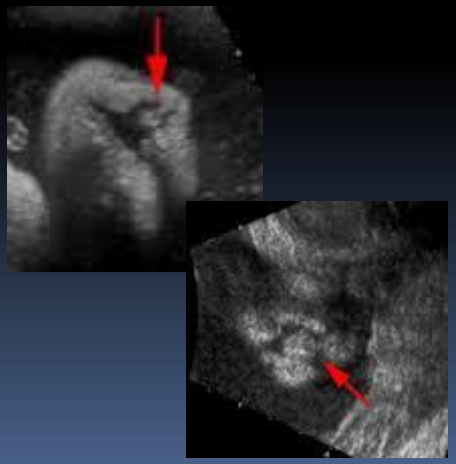


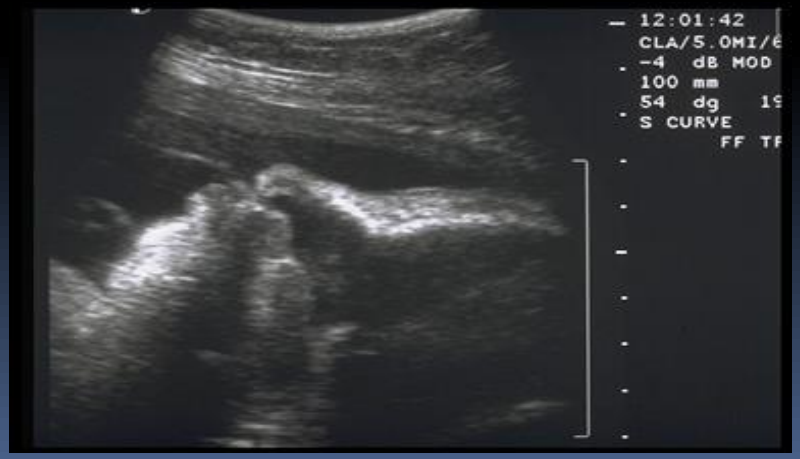
Classificazione

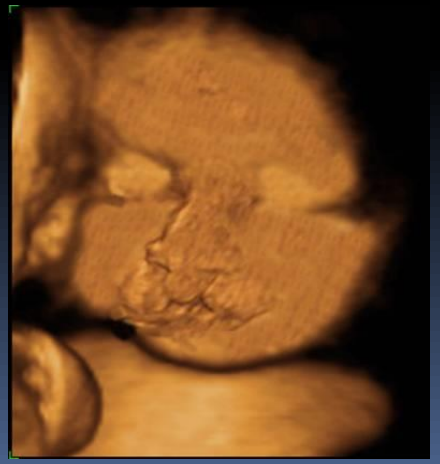


Tipo 3

- Bilaterale
- Scansione coronale, trasversa e sagittale
- Protrusione premascellare
- La protrusione solitamente è più prominente del difetto adiacente
- Nel 59% associato ad aneuploidie, nel 72% al altre anomalie



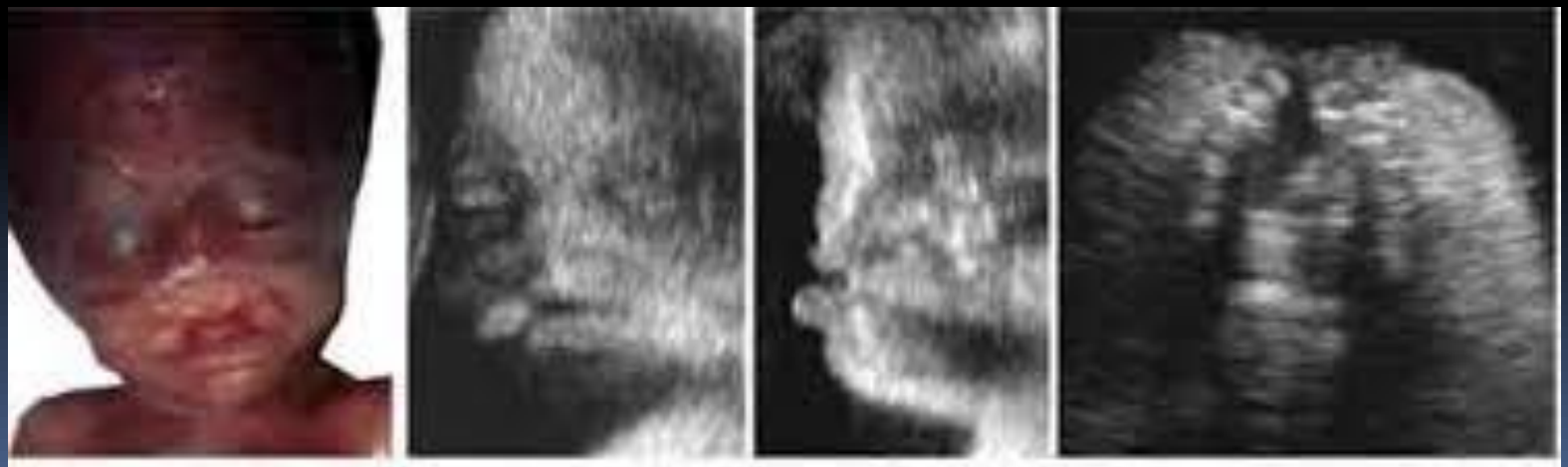




Classificazione

Tipo 4

- Difetto della linea mediana
- Scansioni coronali, trasverse e sagittali
- Ipotelorismo ➡ oloprosencefalia, ipertelorismo ➡ displasia fronto-nasale
- Nel 92% associato ad aneuploidie
- Nel 100% associato ad altre anomalie

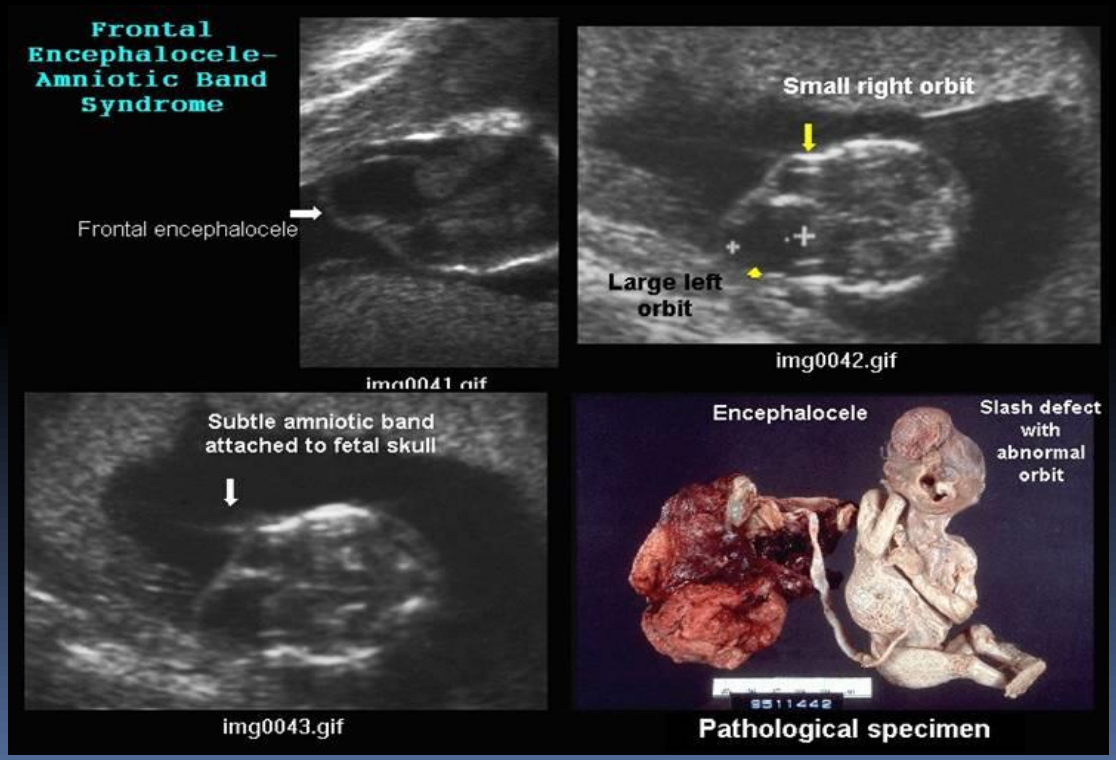


median cleft lip/palate, flat nose, hypothelorism
HOLOPROSENCEPHALY

Classificazione

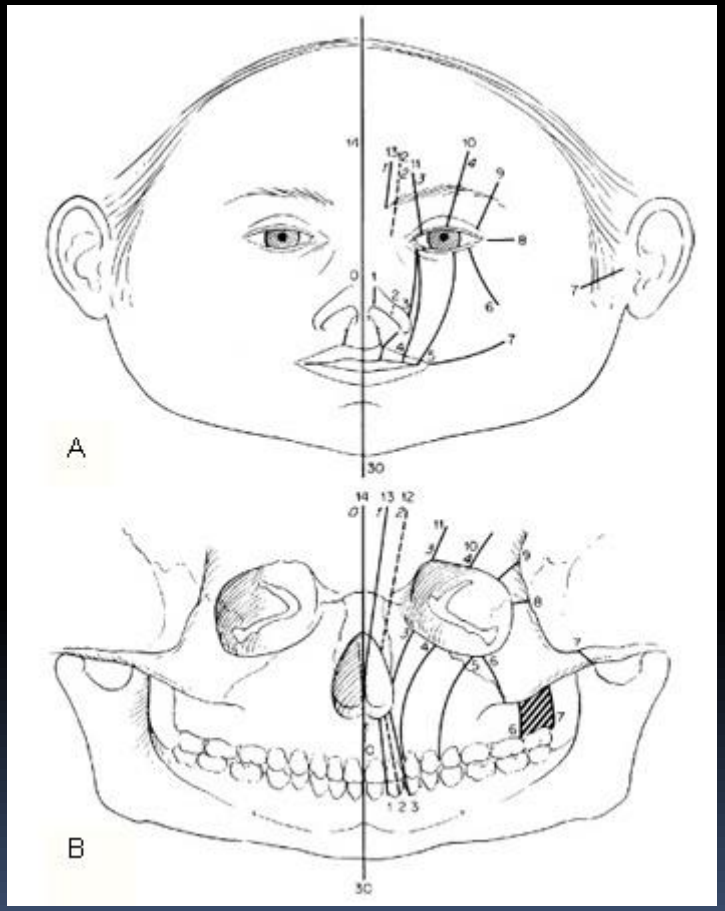
Tipo 5

- Sindrome da banda amniotica
- Solitamente non associato ad anomalie cromosomiche
- Mortalità del 100%



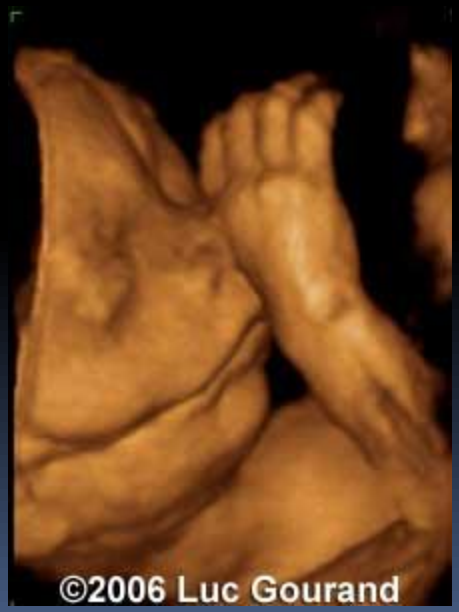
Classificazione di Tessier

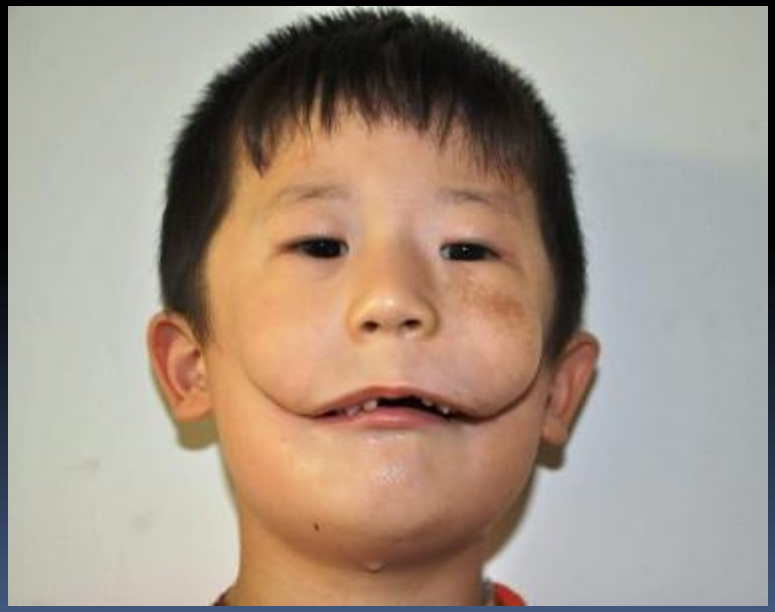
- La maggior parte delle schisi riguarda la giunzione tra premascella e mascella laterale
- Altre varietà sono possibili, ma più rare



Schisi di Tessier n. 7

- Mono o bilaterale
- Ipoplasia dei tessuti ossei (osso zigomatico, mascellare, mandibolare)
- Difficile diagnosi prenatale, utile Eco 3D





Ultrasound Obstet Gynecol 2005; **25**: 12–18
Published online in Wiley InterScience (www.interscience.wiley.com). **DOI:** 10.1002/uog.1819

Ultrasound antenatal diagnosis of cleft palate by a new technique: the 3D 'reverse face' view

S. CAMPBELL*, C. LEES†, G. MOSCOSO‡ and P. HALL§

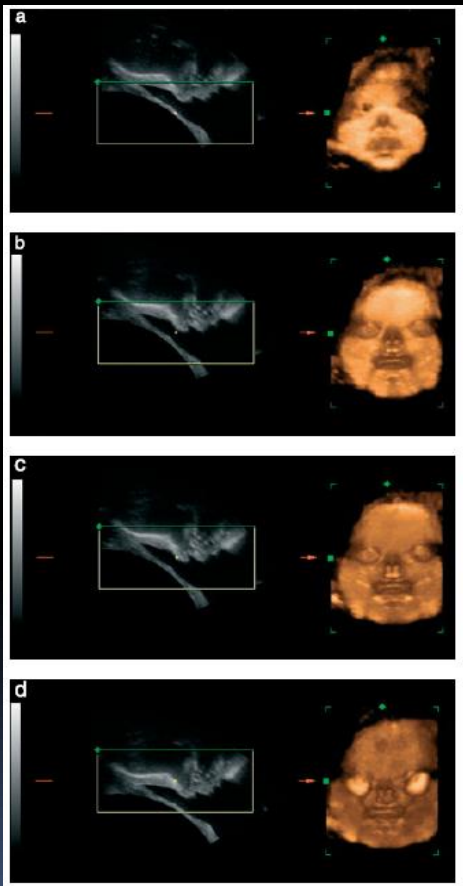


Figure 4 Images corresponding to those in Figure 3 showing the alveolar ridge and palate using the reverse face view. Clear visualization of the orbits, nasal cavity with septum and palate are identified at all levels from the alveolus (a) to the posterior aspect of the hard palate (b–d).

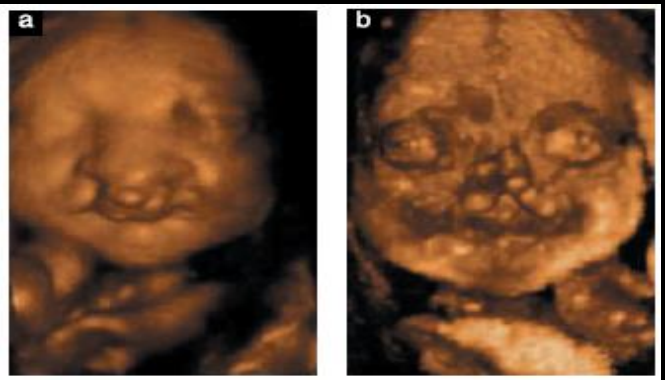


Figure 6 Ultrasound images of Case 1. (a) Frontal view demonstrating bilateral cleft lip. (b) Reverse face view demonstrating cleft in the secondary palate; the fetal tongue has a corrugated echogenic appearance and obscures the margins of the cleft.

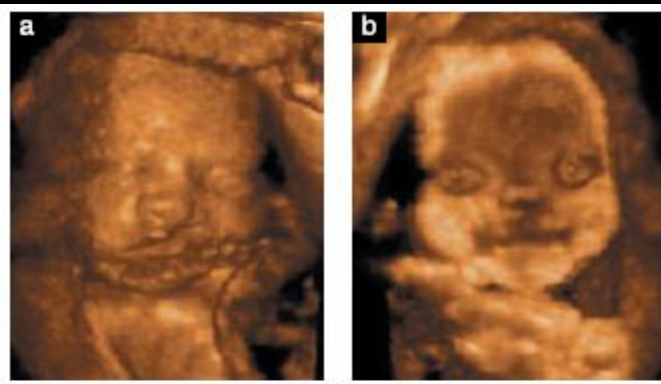


Figure 8 Ultrasound images of Case 3. (a) Frontal view showing left-sided cleft lip; the umbilical cord crosses the chin and left cheek. (b) Reverse face view showing a narrow cleft of the secondary palate.

Methods *Eight cases of suspected orofacial clefting were examined by 3D surface rendering. The fetal lips and alveolar ridge were examined in the frontal plane and the face was then rotated through 180° on the vertical axis to examine the secondary palate by the 3D RF view.*

The 3D technique described here specifically overcomes the shadowing by rotating the frontal facial image through 180° along the vertical axis, so that the palate, nasal cavity and orbits can be approached from the reverse side. Why shadowing occurs when the viewbar is moved

Improving Cleft Palate/Cleft Lip Antenatal Diagnosis by 3-Dimensional Sonography

The "Flipped Face" View

Lawrence D. Platt, MD, Greggory R. DeVore, MD, Dolores H. Pretorius, MD

involving the palate. ***Methods.*** The fetal face was initially examined with the fetus in the supine position. Using 3-dimensional sonography, a static volume was acquired. Following acquisition of the volume, it was rotated 90° so that the cut plane was directed in a plane from the chin to the nose. The volume cut plane was then scrolled from the chin to the nose to examine in sequential order the lower lip, mandible, and alveolar ridge; tongue; upper lip, maxilla, and alveolar ridge; and hard and soft palates. ***Results.*** This approach identified the full length and width of the structures of the mouth and

Figure 4. **A**, Profile of the fetal face. The acquired image is shown in **a**. The purple circle illustrates the reference point, which is placed at the level just below the philtrum. The acquired multiplanar images perpendicular to this point are displayed in **b** and **c**. The rendered image of the face is displayed in **d**. The white arrows illustrate the direction that the image will be rotated. **B**, Change in images as the image in ***a*** is rotated (white arrows). The rendered chin is now observed. **C**, Further rotation in plane ***a*** with the rendered image now looking from the chin toward the nose. **D**, Completion of the rotation with the green cut plane looking at the level of the tongue. This is the final orientation of the green cut plane used to display the rendered image for evaluation of the lips, alveolar ridges, mandible, maxilla, and hard and soft palates.

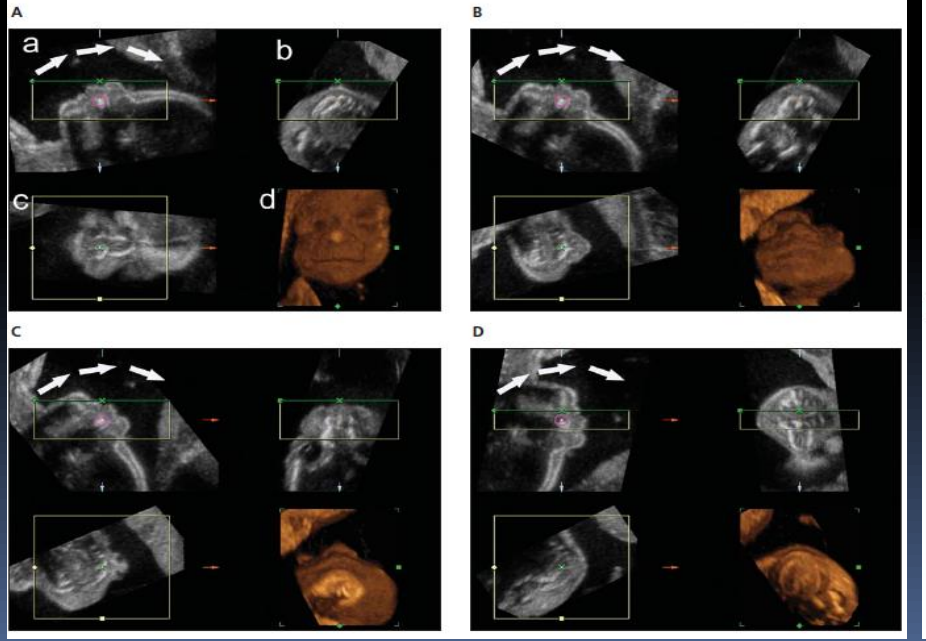
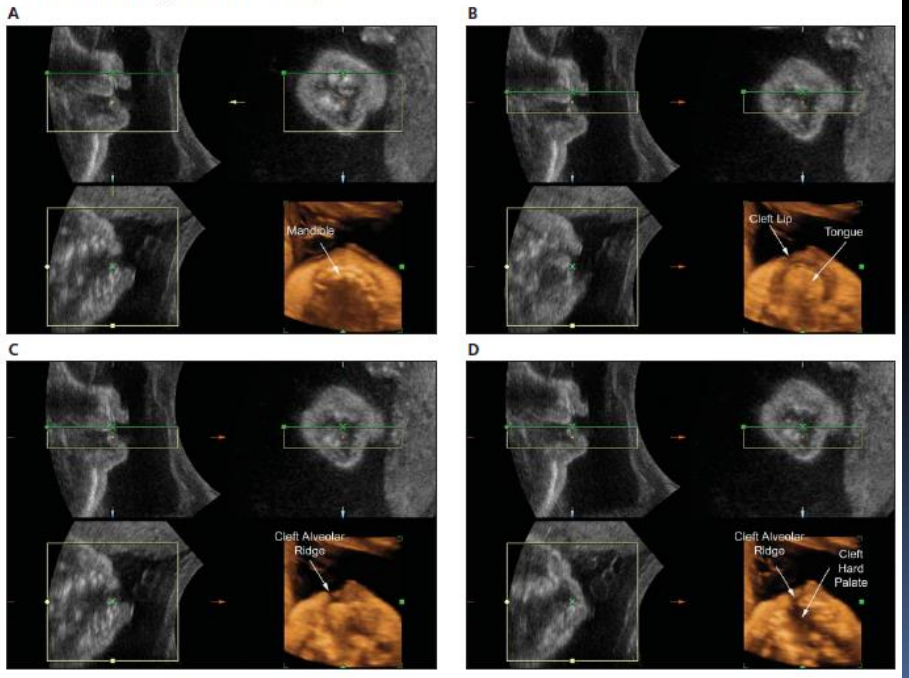


Figure 7. After completing the rotation of the image described in Figure 6, the green cut bar is directed from the lower chin toward the nose to identify the cleft lip and cleft palate. **A**, Mandible and alveolar ridges; **B**, cleft lip and tongue; **C**, cleft of the alveolar ridge and lip; and **D**, continuity between the cleft of the alveolar ridge and the hard and soft palates.



Ultrasound Obstet Gynecol 2007; **29**: 166–169
Published online 17 November 2006 in Wiley InterScience (www.interscience.wiley.com). **DOI:** 10.1002/uog.3877

A novel technique for visualization of the normal and cleft fetal secondary palate: angled insonation and three-dimensional ultrasound

G. PILU and M. SEGATA

Methods *Sonographic examinations were performed in normal fetuses and in one fetus with cleft lip and palate. To avoid acoustic shadowing from the alveolar ridge, the secondary palate was insonated at a 45° angle in the sagittal plane, and 3D ultrasound was used to reconstruct axial and coronal planes.*

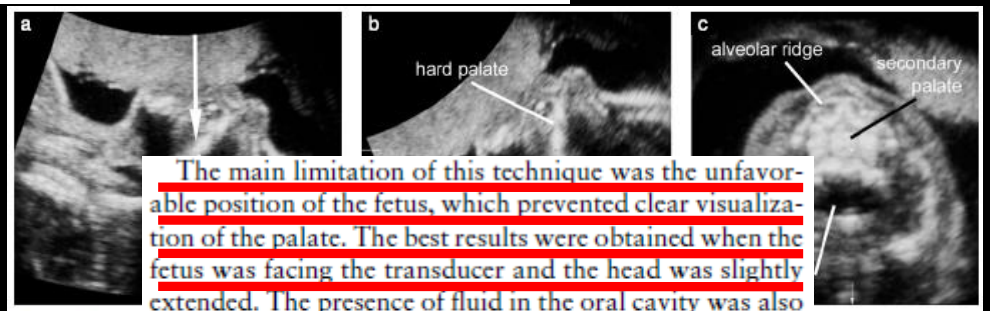


Figure 1 The differ… midsagittal view o… (b) A static three-d… in multiplanar mo… the sagittal section … om the alveolar ridge a … e of about 45° (arrow). c) Reslicing the volume … te can be appreciated in

The main limitation of this technique was the unfavorable position of the fetus, which prevented clear visualization of the palate. The best results were obtained when the fetus was facing the transducer and the head was slightly extended. The presence of fluid in the oral cavity was also a facilitating factor. Part of the soft palate could usually be seen in the sagittal plane but it was not clearly demonstrated in the reconstructed axial and coronal planes.

In conclusion, we propose a new approach to the evaluation of facial anatomy that has the potential to demonstrate the extension of facial clefts into the secondary palate. We believe that this approach may prove useful for demonstrating cleft palate, an anomaly that is not usually amenable to prenatal diagnosis with standard sonographic techniques. However, it is not certain whether this technique would be helpful in diagnosing isolated clefts of the soft palate.

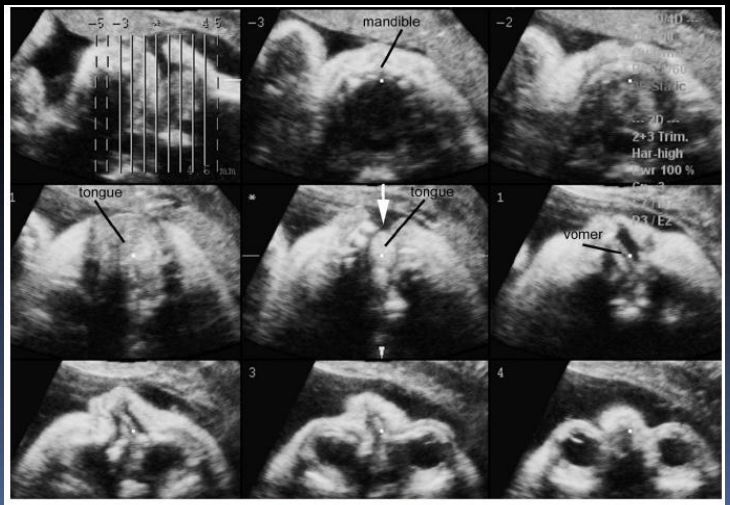


Figure 3 Tomographic ultrasound images in the axial plane of the fetus with cleft lip and palate. The defect of the alveolar ridge is well demonstrated (arrow), but the tongue obscures the edges of the defect. Note the deviation of the nose and of the vomer in the nasal fossae.

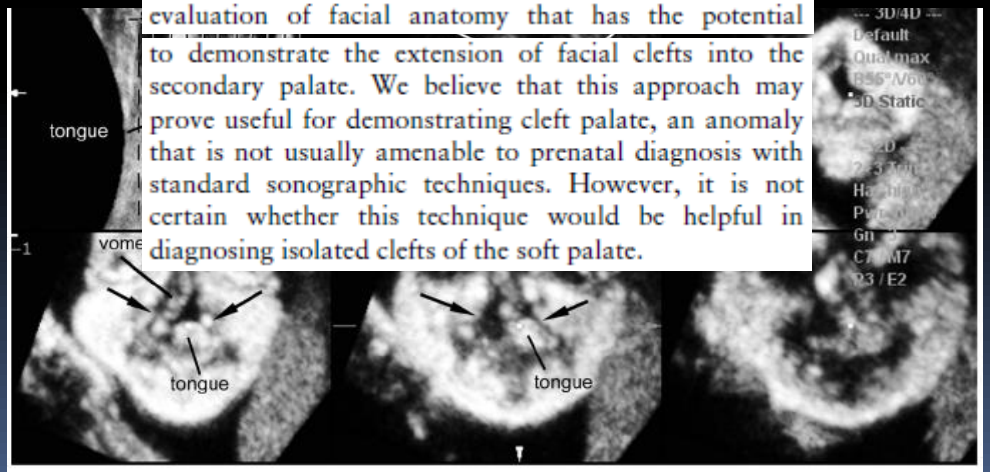


Figure 4 Tomographic ultrasound images in the coronal plane obtained from the same ultrasound volume as that in Figure 3. A comparison between the reference sagittal view and the coronal slices allows discrimination of the tongue from the surrounding tissues. The edges of the defect are well demonstrated (arrows).

Ultrasound Obstet Gynecol 2009; **33**: 399–406
Published online 24 December 2008 in Wiley InterScience (www.interscience.wiley.com). **DOI:** 10.1002/uog.6257

Three-dimensional ultrasound diagnosis of cleft palate: 'reverse face', 'flipped face' or 'oblique face'–which method is best?

P. MARTÍNEZ TEN*, J. PÉREZ PEDREGOSA*, B. SANTACRUZ*, B. ADIEGO*, E. BARRÓN* and W. SEPÚLVEDA†

Methods *A total of 60 fetuses (10 with CLP) with a gestational age ranging from 20 to 33 weeks were examined. We compared visualization of the secondary palate with the previously described reverse-face and flipped-face methods (the latter modified by us) and the oblique-face method developed by us using Oblique View® imaging technology.*

Results *Among the 10 fetuses with CLP the defect involved the lip, alveolus and secondary palate in seven, and the primary palate only in the remaining three. The upper lip and alveolar ridge were well visualized in all cases with all three methods. Involvement of the hard palate was diagnosed correctly in 71% (5/7) of the cases using the reverse-face view, in 86% (6/7) with the flipped-face view, and in 100% (7/7) with the oblique-face view; the hard palate was correctly found to be intact in 78%, 84% and 86%, respectively, of the 50 normal fetuses examined with each view. Involvement of the soft palate was diagnosed correctly in only one of the seven fetuses with defects of the secondary palate in flipped-face and oblique-face views, and was correctly considered intact in only 16% of normal fetuses in flipped-face view and in 26% in oblique-face images.*

Conclusions *Accurate visualization of the soft palate requires an excellent initially acquired volume, fluid between the fetal tongue and palate, and curving of the plane to follow the structure of the palate. The oblique-face or flipped-face views make it possible to visualize the soft palate well in selected cases.*

Table 1 Percentage of fetuses with cleft lip and palate ($n = 10$) in which abnormal findings were well visualized using each technique

Feature	*Reverse-face view (% (n))*	*Flipped-face view (% (n))*	*Oblique-face view (% (n))*
Lip	100 (10/10)	100 (10/10)	100 (10/10)
Alveolar ridge	100 (9/9)	100 (9/9)	100 (9/9)
Hard palate	71.4 (5/7)	85.7 (6/7)	100 (7/7)
Soft palate	0 (0/7)	14 (1/7)	14 (1/7)

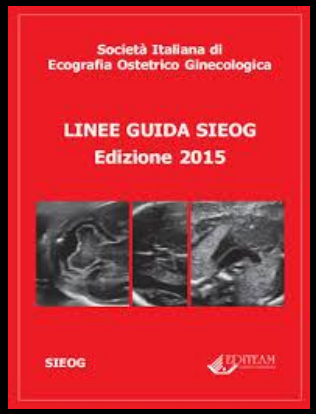
Table 2 Percentage of fetuses without cleft lip or palate ($n = 50$) in which different structures were well visualized using each technique

Feature	*Reverse-face view (% (n))*	*Flipped-face view (% (n))*	*Oblique-face view (% (n))*
Lip	100 (50/50)	100 (50/50)	100 (50/50)
Alveolar ridge	100 (50/50)	100 (50/50)	100 (50/50)
Hard palate	78 (39/50)	84 (42/50)	86 (43/50)
Soft palate	0 (0/50)	16 (8/50)	26 (13/50)

Ruolo della RM

LINEE GUIDA

RISONANZA MAGNETICA FETALE INDICAZIONI, PROTOCOLLI DI STUDIO E SICUREZZA

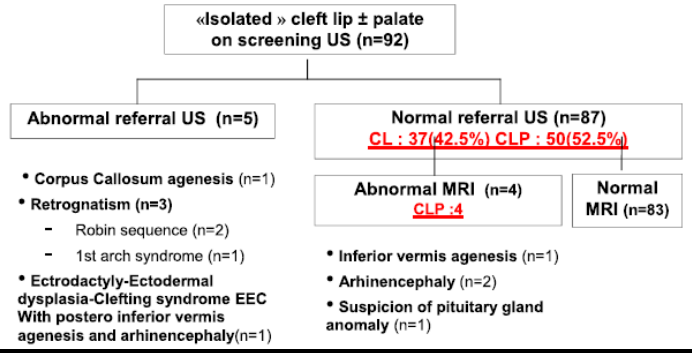


f. Malformazioni del distretto cranio-facciale in particolare la labiopalatoschisi bilaterale. La labiopalatoschisi può essere isolata o rientrare nel contesto di sindromi genetiche; circa l'11% dei pazienti affetti da schisi labiale, infatti, può essere inquadrato nel contesto di una delle oltre 170 sindromi monogeniche riportate nel London Dysmorphology Database e il 25-30% dei neonati affetti da malformazioni del massiccio facciale presenta patologie associate, quadri sindromici o aneuploidie (34). Lo studio 3D ottiene in condizioni favorevoli di posizione fetale e di quantità adeguata di liquido amniotico una buona definizione della patologia. Tuttavia la RM, nei casi di labiopalatoschisi bilaterale in considerazione dell'alta associazione con quadri sindromici e anomale del SNC è in grado di aggiungere particolari dettagli sulle anomalie intracraniche (35, 36).

Is fetal cerebral MRI worthwhile in antenatally diagnosed isolated cleft lip with or without palate?

Marie-Noëlle de Milly[1]*, Marianne Alison[1], Owen Arthurs[1], Edith Vuillard[2], Jean-Francois Oury[2], Monique Elmaleh-Berges[1], Guy Sebag[1] and Nadia Belarbi[1]

Table 1 Summary of patients included in this study



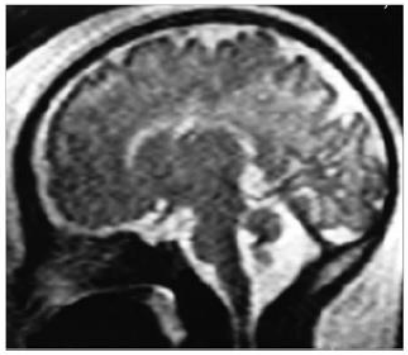


Figure 1 Inferior vermis agenesis prenatal magnetic resonance imaging performed at 33 weeks of gestation midline sagittal T2-weighted sequence demonstrating the small size and the abnormal orientation of the vermis

WHAT'S ALREADY KNOWN ABOUT THIS TOPIC?

- Cleft lip/palate may be isolated or associated with other malformations that must be looked for antenatally. The most common associated anomalies affect the limbs and spine, the cardiovascular system, and the central nervous system.

WHAT DOES THIS STUDY ADD?

- The incidence of associated cerebral abnormalities detected on MRI in isolated cleft lip/palate is 4.6%. Ultrasound may be sufficient for their detection. MRI is useful if the ultrasound examination is technically difficult.

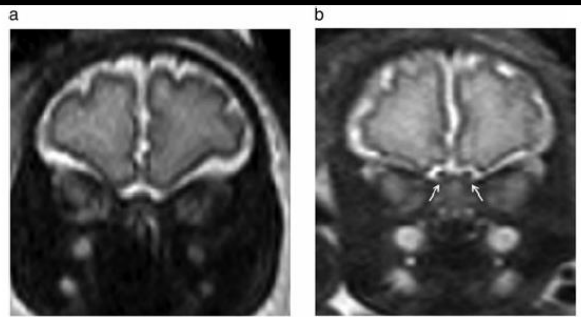


Figure 2 Arhinencephaly prenatal magnetic resonance imaging at 32 weeks of gestation: coronal T2-weighted sequences. a: Absence of the olfactory bulbs. b: Normal fetus for comparison: normal aspect of the olfactory bulbs (arrows)

Prenatal Diagnosis 2013, 33, 273–278

Management ostetrico

- *Ecografia* 2D-3D per indagare e classificare correttamente il difetto e ricercare eventuali altre anomalie associate
- Adeguato *counselling*
- *Cariotipo*
- Valutare eventuale *RM*
- *Counselling chirurgico* (tempi e modalità, possibili risultati funzionali ed estetici del trattamento, disturbi fonazione, udito e respirazione)