

Conclusion

The consistent and systematic exploitation of the 2D ultrasound capabilities allows the comprehensive diagnosis of fetal facial clefts and of the extent of CLP, including palate involvement. This had previously been a major challenge. The systematic exploration of the fetal pharyngeal space focusing on finding and depicting the uvula allows the investigator to detect an indication of isolated cleft palate which previously had been viewed as essentially undiagnosable. An unremarkable uvula, which can be depicted as a typical "equals sign", rules out isolated cleft palate. If the uvula cannot be detected, the soft palate must be depicted in sagittal view. If the soft palate cannot be depicted, isolated cleft palate can also be diagnosed with certainty.

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2D Sonographic Evaluation of Fetal Facial Clefts

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Introduction

With an incidence of about 1:1000 births, facial clefts are among the most frequently occurring congenital malformations^{1,2}. They can take on various forms, but from a pathophysiological point of view cleft lip and palate (CLP) significantly differ from isolated cleft palate (CP). In CLP the defect always starts at the lip and extends to a variable degree in a dorsal direction (lip notch, cleft lip, CLP (hard palate only or hard and soft palate)). Isolated cleft palate always starts at the uvula (uvula bifida as the mildest form) and to a variable extent proceeds along the midline in anterior direction. It either affects only the soft palate or the soft and hard palate. Both CLP and CP can occur as isolated malformations or as components of chromosomal or syndromal disorders⁷.

While prenatal detection of CLP during a differentiated ultrasound examination is usually fairly unproblematic, it is significantly more difficult to evaluate the palate structures involved in CLP to assess the extent of the malformation.

The major challenge is the detection of isolated cleft palate. The prenatal detection rate of isolated cleft palate approaches zero (0–1.4%)^{3–8} which shows that obviously at present there are no satisfactory sonographic indicators available to confirm the existence of isolated cleft palate. Because of its dome-shaped structure, the palate cannot be visualized in its entirety in 2D. Moreover, the surrounding osseous structures further complicate the evaluation due to imaging artifacts in the region of interest.

Presenting different case studies, this paper will not only show that CLP and CP can be diagnosed in 2D but also how to perform this examination.

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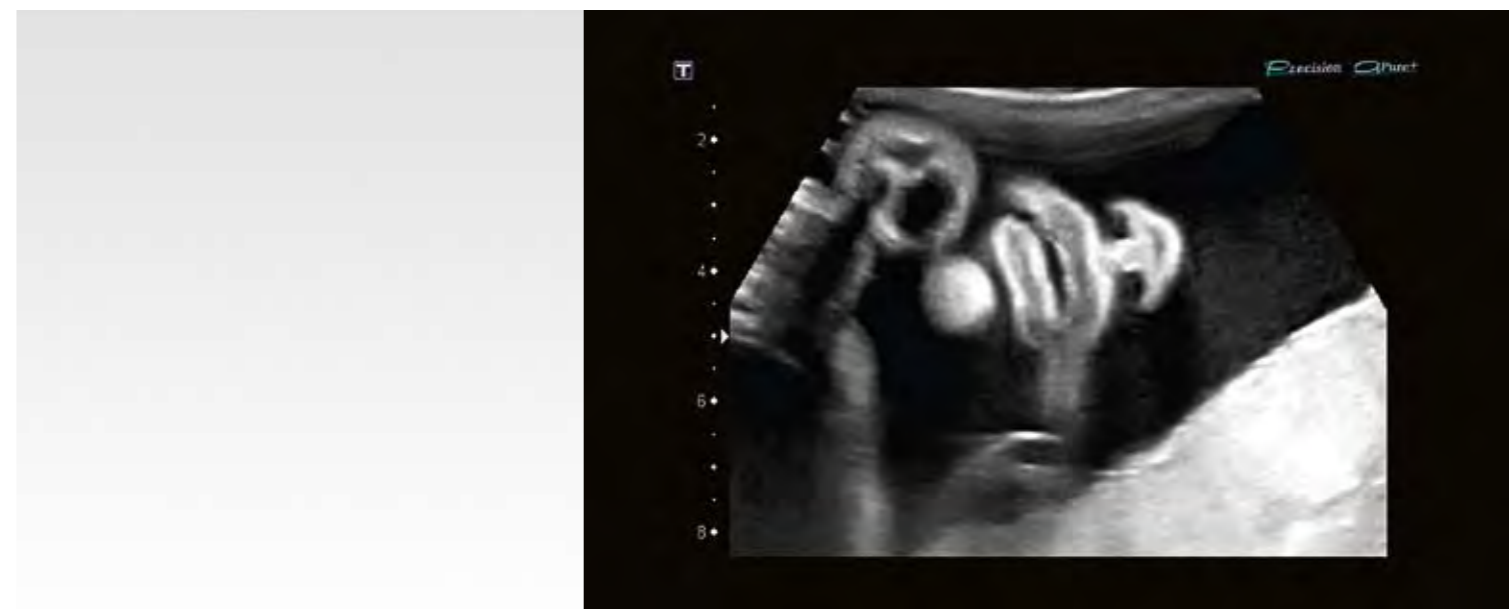


Fig. 1: Frontal facial view with depiction of lips, nose with nostrils and chin.

Case Studies

1) Normal Findings

Lips are best evaluated in a frontal view with depiction of the upper lip, lower lip and nose with nostrils (Fig. 1), which in most cases allows confirmation of the integrity of the upper lip.

The integrity of the maxilla is evaluated in the axial view at the level of the upper jaw which also allows

checking the continuity of the upper lip (Fig. 2). The hard palate, however, cannot be depicted in this way, since it is partly out of plane and ultrasound artifacts prevent an evaluation behind the bone.

Because direct and complete evaluation of the entire palate is impossible, the relevant parts of the palate have to serve as reference points while the typical pathophysiology of the different types of clefts has

to be taken into consideration. Confirmation of an intact upper lip rules out a cleft palate in CLP. An unremarkable uvula rules out isolated cleft palate, because with isolated cleft palate the uvula is the structure which is always affected (with the exception of submucous clefts). Clefts in the area of the soft palate are situated along the midline in both CLP and CP. Both the depiction of the uvula in frontal or transversal view through the pharynx (Fig. 3 and 4)

as the typical “equals sign” and the depiction of the soft palate in the median sagittal view (Fig. 5) succeed very often and thus facilitate ruling out isolated cleft palate⁹. If in these cases the upper lip is intact, the hard palate must also be intact. Visualization of the hard palate in sagittal view is more difficult because of the ultrasound angle and only succeeds in fetuses with the neck extended (Fig. 6).

2) Isolated Cleft Lip

In the frontal view at the level of the lips, the defect on the left side of the upper lip can be depicted (Fig. 7). The next structure to be evaluated is the maxilla which can easily be visualized in axial view without any loss of continuity (Fig. 8). Both the uvula (as a typical “equals sign”, Fig. 9) and the entire palate in sagittal view (Fig. 10) are unremarkable. Thus, in this case the cleft lip can be

confirmed without any involvement of the jaw and the palate. Figure 11 shows the postnatal image of this lesion.



Fig. 2: Axial view at the level of the upper jaw with depiction of the unremarkable maxilla (↓).



Fig. 3: Frontal view of the neck with depiction of the uvula (o, “equals sign”). The key structure to find the uvula in this view is the epiglottis (↓).



Fig. 7: Depiction of a lip defect on the left side in frontal view (↓).



Fig. 8: Axial view at the level of the upper jaw with confirmation of an intact maxilla (o).



Fig. 4: Depiction of the uvula (o) as a typical “equals sign” in a tilted, more posterior transverse view at the level of the base of the skull.



Fig. 5: Depiction of the soft palate (↓) with uvula (o) in a median sagittal view.



Fig. 9: Unremarkable uvula (o) in a frontal view through the pharynx.



Fig. 10: Depiction of an intact palate (↓) with uvula (o) in a median sagittal view.



Fig. 6: Depiction of the entire palate in a median sagittal view with neck extended. Hard palate (↓), soft palate (↓) with uvula (o).

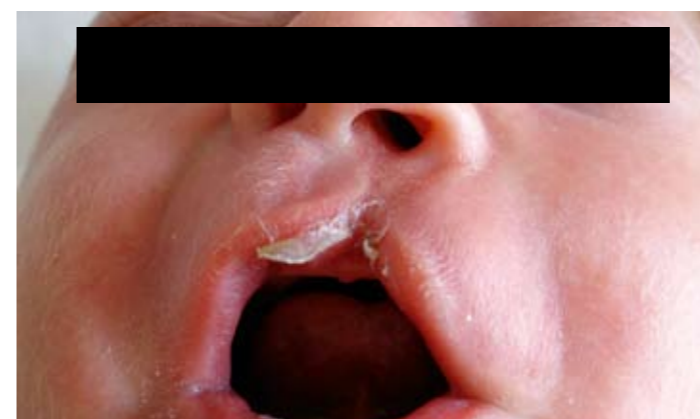


Fig. 11: Postpartum image with confirmation of the isolated lip defect.

3) Unilateral CLP

In frontal view a large defect of the upper lips on the right side can be easily visualized (Fig. 12). The axial view at the level of the upper jaw shows the continuity loss of the maxilla and confirms the involvement of the jaw (Fig. 13). The absence of

visible palate structures in the median sagittal view confirms palate involvement (Fig. 14). This is supported by the fact that the uvula cannot be detected (Fig. 15). The postpartum image confirms the prenatal diagnosis (Fig. 16).



Fig. 12: Depiction of an extensive lip defect (↓) on the right side in a frontal view.



Fig. 13: Depiction of a discontinuity in the maxilla (↓) in axial view at the level of the upper jaw.



Fig. 14: Detection of the absence of palate structures including the uvula (↓) in a median sagittal view.



Fig. 15: Detection of the absence of the uvula in a frontal view through the pharynx (○); epiglottis (↓).



Fig. 16: Postpartum image with confirmation of the prenatal diagnosis. The visible clip (↓) is not a palate structure, but a device used for orthodontic therapy.

4) Unilateral CLP with Detection of a Split Uvula

The frontal view shows a cleft in the region of the left upper lip (Fig. 17) which is not as extensive as in the previous case. Moreover, the defect in the maxilla, which is seen in the axial view, is not very

extensive (Fig. 18). The anterior portion of the soft palate cannot be depicted in median sagittal view, but an uvula can be seen (Fig. 19). The examination of the uvula in frontal view through the pharynx shows a completely split but very closely spaced uvula (double "equals sign", Fig. 20). The diagnosis

was confirmed after birth, but postpartum images for this child were regrettably unobtainable.



Fig. 17: Depiction of lip involvement (↓) in a frontal view with CLP on the left side.



Fig. 18: Detection of the maxillary defect (↓) in axial view.



Fig. 19: In a median sagittal view only one palate structure can be depicted that stands out as an uvula (○). Otherwise, it is not possible to visualize a portion of the soft or the hard palate (↓).

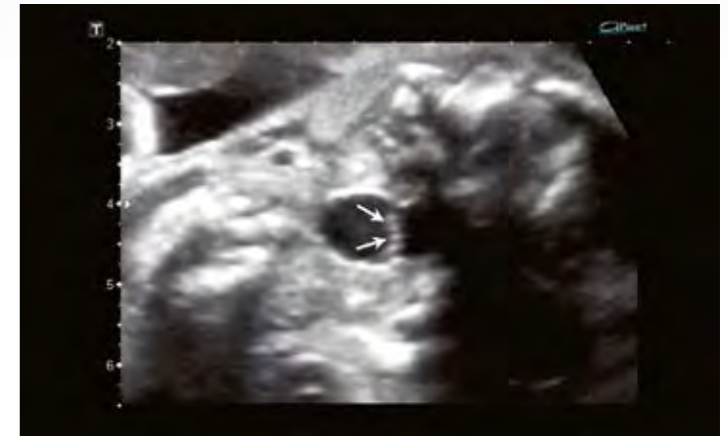


Fig. 20: In a frontal pharyngeal view the split uvula is detected as double "equals sign". This confirms the involvement of all structures of the palate. In the region of the soft palate the cleft is only very narrow.

5) Bilateral CLP

The frontal view shows a bilateral cleft of the upper lip. The defect is more pronounced on the right side than on the left side (Fig. 21). In the axial view the right-side defect of the maxilla can be depicted while the continuity on the left side is

completely preserved (Fig. 22). The median sagittal view with absent depiction of soft palate structures confirms the involvement of the entire palate (Fig. 23). This is supported by the inability to detect the uvula in the frontal view through the pharynx (Fig. 24). The postpartum image confirms the

prenatal diagnosis of CLP with bilateral lip involvement as well as jaw and complete palate involvement on the right side (Fig. 25).



Fig. 21: Depiction of a bilateral lip defect (↓), on the right more extensive than on the left.



Fig. 22: Axial view with detection of maxilla involvement on the right (↓), while the maxilla on the left side is depicted without any loss of continuity (○).



Fig. 23: Detection of the absence of palate structures in a median sagittal view (↓). This shows the involvement of the entire palate in this CLP.



Fig. 24: Detection of the absence of the uvula (○) above the epiglottis (↓).



Fig. 25: Postpartum image which confirms the prenatal diagnosis.

6) Isolated Cleft Palate (CP)

The frontal view of the face shows the integrity of the upper lip but no cleft (Fig. 26) while the exploration of the pharynx does not show an uvula (Fig. 27) which indicates isolated cleft palate. In median sagittal view neither parts of the soft palate

nor the hard palate can be depicted (Fig. 28) which points at a more extensive isolated cleft palate. This was confirmed postpartum (Fig. 29).



Fig. 26: Frontal view with depiction of an unremarkable oral-nasal region.



Fig. 27: While examining the pharyngeal space, the uvula (○) cannot be visualized above the epiglottis (↓).



Fig. 28: Detection of the absence of the palate structures in a median sagittal view (↓) resulting in the certain diagnosis of an isolated cleft palate.



Fig. 29: Postnatal image with depiction of the isolated cleft palate (hard and soft palate).