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Facial cleft detected: Is the palate normal?

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Despite advances in ultrasound technology, the sensitivity for detection of facial clefts at the routine mid-trimester details scan remains relatively poor. This can be improved by the use of a three-point ultrasound screening protocol, although this is not routine in many countries. When a facial cleft is suspected at the routine scan, further imaging is usually required to detail the extent of the cleft and presence or absence of any other abnormalities. Involvement of the fetal palate is an important finding that will determine the requirement for surgery, audiology, and orthodontic services well into teenage years. There remains little uniformity in how a facial cleft is described antenatally, with involvement of the alveolar ridge frequently and incorrectly taken to mean involvement of the palate. Further, midline clefts of the hard and soft palates, where the fetal lips and alveolar ridge are intact, are a feature of many genetic conditions, but are almost never diagnosed by prenatal ultrasound. In this chapter, we detail issues surrounding the nomenclature of facial clefts in relation to the palate, and describe some of the more commonly used two-dimensional and three-dimensional methodologies for imaging the fetal palate.

Introduction

Prenatal detection rates for isolated clefts of the lip and alveolus by two-dimensional ultrasound have historically been poor, ranging from 45–68% [1–3], although a more focused approach using high-resolution equipment and additional scanning planes ('face on'; coronal plane through the alveolar

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ridge and sagittal profile view) has resulted in an improvement from 34–58% in two consecutive time epochs through the introduction of the additional scanning planes detailed [4].

One of the most recent papers on cleft detection, from Southern Sweden reflecting contemporary practice in the period 2006–2010 [5], details a somewhat disappointing detection rate of 31% (43% if isolated midline clefts are excluded). This detection rate compares unfavourably with a study that most closely resembles UK practice, based on birth registry data from 2005–2006. This reported a 64% rate of prenatal detection for cleft lip, palate, or both [6]. It is possible that the detection for facial clefts has fallen in the UK since the recommendation in January 2010 that the 'face on' view only should be carried out as part of the anomaly scan [7].

Although there is little doubt that the use of higher resolution ultrasound equipment and incorporation of protocols for examining the fetal face at the time of the detailed anomaly scan can improve sensitivity to as high as 75%, the detection rates for clefts of the secondary (hard) palate using two-dimensional ultrasound have remained low, even in those patients in whom a cleft lip has been detected. Secondary palatal involvement, however, can alter the long-term prognosis for the child, being more commonly associated with difficulties in speech, hearing and (after surgery) mid-facial protrusion. Typically, clefts of the secondary palate require more corrective surgery into teenage years, and a greater input of audiology, speech and language therapy, and orthodontic intervention. The accuracy of antenatal diagnosis of facial cleft is, therefore, particularly helpful in preparing parents and cleft teams for the birth of a child with facial cleft, and for advance planning.

Recent years have seen improvements in two-dimensional imaging and the advent of three- and four-dimensional ultrasound technology, coupled with different techniques for visualising the fetal face and, in particular, the hard and soft palates. Little uniformity exists in approach between specialists in cleft imaging, however, and the many different ways that facial clefts are described. In this chapter, we consider the confusions in nomenclature associated with facial clefts, and the different techniques and ultrasound modalities described to assess the fetal palate, particularly where a facial cleft is suspected.

Embryology

Facial clefts result from defective fusion of the facial processes. Clefts of the lip and alveolus occur because of a failure of fusion of the medial nasal process and maxillary swellings [8]. They may be unilateral or bilateral, and associated with a cleft of the alveolus on the side of the lip cleft. Clefting of the secondary (hard) palate has a distinct and different embryological derivation, and is secondary to a failure of fusion of the two palatine shelves [9].

Clefts of the secondary (hard) palate are, therefore, always midline and posterior to the incisive foramen and, if they occur together with cleft of the lip and alveolar ridge, they are termed unilateral or bilateral cleft lip with cleft palate [9,10]. A cleft of the secondary palate where there is also a cleft lip cannot occur in the absence of cleft of the ipsilateral alveolar ridge except for the rare situation of a baby suffering from both embryological variants of facial cleft. Facial clefts have been typically divided into cleft lip with or without palatal involvement. Involvement of the lip and alveolus was said to represent a cleft of the primary palate, that area anterior to the incisive foramen, with clefts of the secondary palate being described distinctly. Confusion in terminology means that a child with 'cleft lip and palate' may in fact have a cleft of the primary palate but an intact secondary palate; this represents a cleft alveolus.

The embryological origins of isolated clefts of the secondary palate seems to be distinct from those of clefts of the lip and alveolus [8]. The secondary palate comprises the hard palate (immediately posterior to the incisive foramen) and the soft palate (posterior to the hard palate, containing no bone). Isolated cleft palate is rarer than unilateral cleft palate or bilateral cleft palate; ultrasound studies underestimate the incidence of isolated cleft palate, as this condition is rarely diagnosed by prenatal ultrasound and may even not be identified immediately after delivery.

In Denmark, isolated cleft palate accounts for 25% of the total number of cases of facial clefts [11,12], but surveys in other countries show that the incidence varies; for example, it is over 50% in Northern Ireland and Scotland [13–16]. Secondary palate clefts are always midline and result from the failure of the palatine processes to elevate or grow, so a cleft of the hard palate must also affect the soft palate; however, it is possible for the soft palate to be cleft with the hard palate.