Review Article

Prenatal Ultrasound Imaging of Orofacial Clefts: A Pictorial Essay

CME Credits

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Abstract

Orofacial clefts (OFCs), including cleft lip (CL), cleft palate (CP), and CL with palate (CL/P), are relatively common congenital birth defects occurring in approximately 1 in 500 to 1 in 2500 live births. Detecting OFCs during prenatal ultrasound screening is crucial for informed decision-making and multidisciplinary medical care. This review provides a practical guide for routine and advanced screening for OFCs during mid-pregnancy. The Maarse classification system facilitates effective communication among the multidisciplinary team, categorizing OFCs into five types. Basic ultrasound views encompass coronal, sagittal, and axial imaging of the face and hard palate. Additional visualization techniques are employed in case of suspected anomalies during the initial screening. Advanced ultrasound views provided by the expert in prenatal OFC diagnosis include imaging of the posterior edge of the hard palate and the posterior part of the soft palate. Detected OFCs exhibit a range of severity and affect different structures, underscoring the importance of accurate detection and classification for appropriate treatment planning. Implementing a standardized screening protocol for OFCs is essential. By enhancing detection rates and enabling early diagnosis, prenatal ultrasound screening contributes to improved patient outcomes and facilitates timely intervention by the multidisciplinary team. In conclusion, this review emphasizes the significance of standardized protocols and specialized techniques for prenatal ultrasound screening of OFCs. Early detection and classification of these malformations play a vital role in comprehensive management, ensuring that affected individuals and their families receive the appropriate care and support they need.

Keywords: Cleft lip, cleft palate, hard palate, orofacial cleft, patient care team, ultrasonography prenatal

Introduction

Orofacial clefts (OFCs) constitute a diverse group of congenital birth defects, including cleft lip (CL), cleft palate (CP), and CL with palate (CL/P). They rank among the most common craniofacial congenital malformations, with a global incidence of 1 in 700 individuals.[1] The prevalence of OFCs largely varies between populations, with the highest rates reaching 1 in 500 live births among North American and Asian populations, whereas rates among African and African American populations are as low as 1 in 2500 live births.[2] OFCs can be isolated or coincide with various chromosomal anomalies and phenotypic syndromes, posing physical, psychological, and socioeconomic challenges for both patients and caregivers. Early diagnosis is crucial, providing parents with comprehensive information about the condition and enabling preparation for an extended course of multidisciplinary medical care.[3,4]

Received: 04-10-2023 **Accepted:** 01-11-2023 **Available Online:** 21-03-2024

Access this article online

Quick Response Code:

Website: https://journals.lww.com/jmut

DOI: 10.4103/jmu.jmu_123_23

Ultrasound screening during mid-pregnancy serves two primary goals: first, detecting the presence of OFCs, and second, defining the cleft type based on anatomical markers. This distinction is critical for assessing the necessity of postnatal surgical intervention and referral for multidisciplinary team management. Although prenatal screening for OFCs is a standard of care in numerous countries, challenges persist.[5] Despite improvements in methodology and ultrasound techniques, the diagnosis of OFCs remains challenging due to a lack of standardized protocols, particularly for CP.[6] Moreover, ultrasound examinations can be hindered by the factors such as gestational age, maternal obesity, volume of amniotic fluid, coexisting fetal anomalies, fetal motility, or the experience of the radiological technician.[7] This review aims to consolidate visual clues and practical methods, offering

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How to cite this article: Yang TY, Chang TY. Prenatal ultrasound imaging of orofacial clefts: A pictorial essay. J Med Ultrasound 2024;32:8-13.

an easy-to-follow, step-by-step guide for routine and advanced OFC screening during early or mid-pregnancy gestation.

Prenatal Ultrasound Classification of Orofacial Clefts

As previously mentioned, OFCs constitute a group of malformations that necessitate systematic classification for effective communication within the multidisciplinary team during both the pre- and postnatal periods. In our practice, we employ the typology proposed by Maarse *et al*.,[1] designed to bridge the gap between postnatal and prenatal diagnosis, facilitating multidisciplinary management of OFCs.[8] The classification includes the following types:

- Type 1: CL
- Type 2: CL and alveolus
- Type 3: CL, alveolus, and palate
- Type 4: CP only
- Type 5: Submucous CP.

Types 1 through 3 can be unilateral (u) or bilateral (b), and the involvement of the nostril floor determines if it is complete or incomplete. Isolated CPs (type 4) are often undiagnosed until birth, and submucous CPs (type 5) may go unnoticed even after birth. Figure 1 schematically illustrates these types.

A multidisciplinary team approach is widely applied to the management of OFCs.[4] It aids in the provision of a detailed prenatal diagnosis, counseling parents regarding the condition, and planning surgical and postnatal comprehensive care. In our clinic, based on the experiences and literature review, we follow a step-wise protocol illustrated in Figure 2.

Basic Ultrasound Views for Prenatal Screening of Orofacial Clefts

During routine mid-trimester screening, each fetus undergoes an anatomical scan of the face and hard palate. The basic mid-trimester routine ultrasound involves capturing images from coronal, sagittal, and axial views.[9] To enhance the detection rate for facial anomalies, especially clefts, we will outline the steps followed in our clinic during basic screening, along with the diagnostic signs that indicate anomalies.

First, we initiate an axial plane sweep starting from the level of the eye socket [Figure 3a] and progressing downward to the level of the maxillary alveolus [Figure 3b] and mandible [Figure 3c]. This allows us to assess the alveolus, with the landmark being the entire alveolar ridge, which should appear continuous and symmetrical. Next, we tilt the probe toward the fetal chest in an oblique axial view until we can visualize the posterior edge of the hard palate. This edge appears as an echogenic line without interruption [Figure 3d]. In the third step, we rotate the probe by 90° to obtain a true sagittal view, enabling us to assess the fetal profile [Figure 3e]. For this purpose, we utilize the fetal profile line proposed by de Jong-Pleij *et al*.[10] The fetal profile line connects the anterior border of the mandible with the intersection between the nasal and frontal bones. A normal profile is indicated when the fetal profile line aligns longitudinally with the frontal bone, corresponding to a position of zero. Finally, in the coronal nose-mouth view, we examine the nostrils and upper lip [Figure 3f]. These structures should be intact and symmetrical, and the entire upper lip should be visible. By

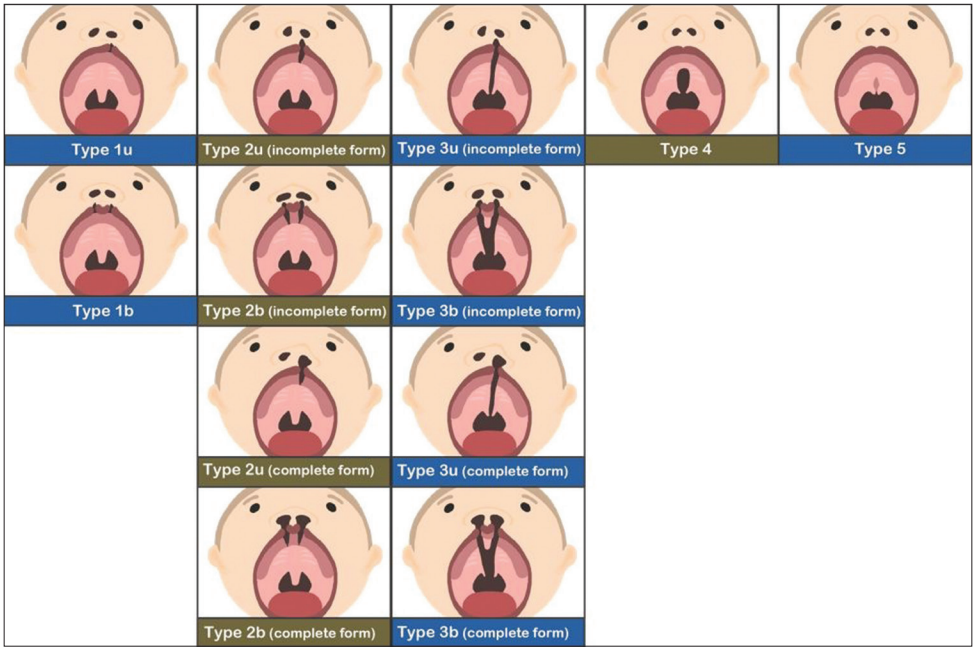

Figure 1: Schematic representation of different orofacial cleft types based on classification by Maarse *et al*.[1] Type 1u: Unilateral cleft lip. Type 2u: Unilateral cleft lip and alveolus. Type 3u: Unilateral cleft lip, alveolus, and palate. Type 1b: Bilateral cleft lip. Type 2b: bilateral cleft lip and alveolus. Type 3b: Bilateral cleft lip, alveolus, and palate. Type 4: Cleft palate only. Type 5: Submucous cleft palate

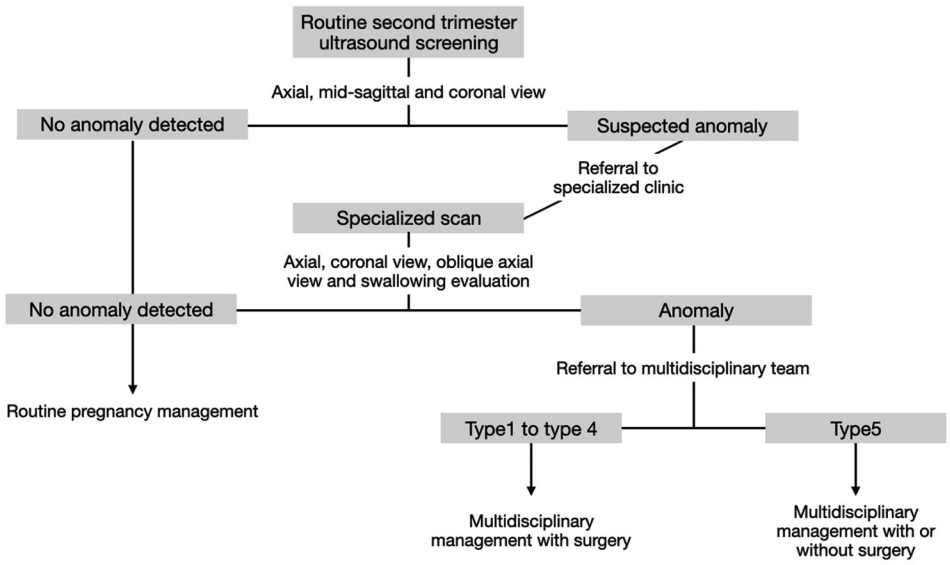

Figure 2: Flow diagram for ultrasound diagnosis and follow-up management for orofacial clefts

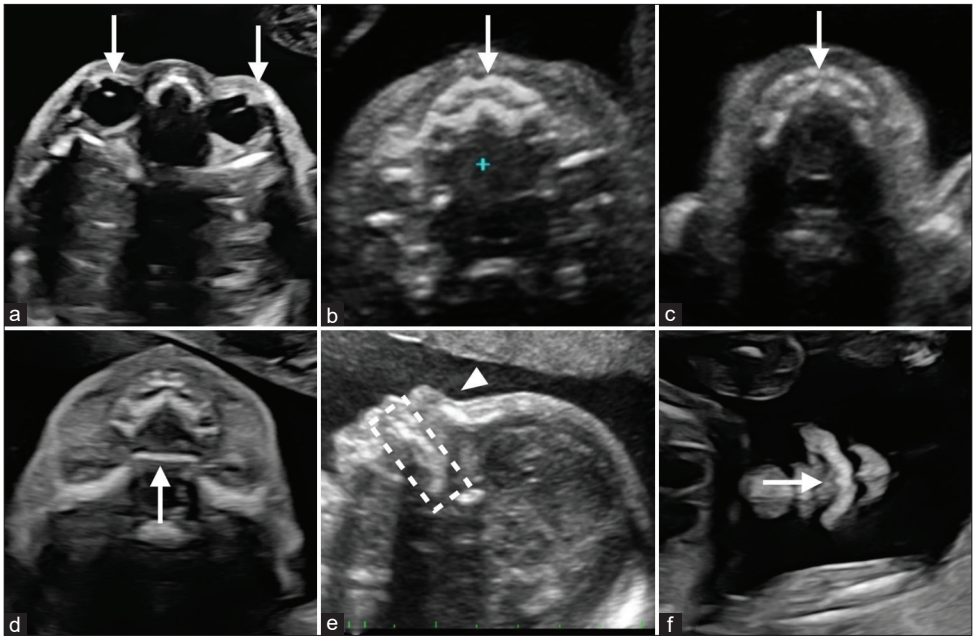

Figure 3: A series of two-dimensional grayscale ultrasound images for basic orofacial cleft (OFC) screening. A stepwise basic screening for OFCs includes visualization of: the eye sockets (arrows, a), maxillary alveolus (arrow, b), mandible (arrow, c), and the posterior edge of the hard palate (arrow, d) in the axial view. Besides the axial view, the facial profile was obtained in true sagittal view to visualize the nasal bones (arrowhead, e), and maxillary alveolus (dashed rectangle, e). Finally, the fetal lip was imaged in the coronal view (arrow, f)

following these steps and observing abnormal findings on the described planes, our goal is to identify cases suspected of OFCs and refer them for advanced screening by a specialist during the mid-trimester screening.

Advanced Ultrasound Views for Prenatal Screening for Orofacial Clefts

Advanced screening is recommended in cases where an anomaly is suspected during basic screening. All scans in the specialized clinic are performed by the fetal-maternal medicine doctor with expertise in OFCs. The key views are identical to the basic screening: the lips and the maxillary alveolus. The specialized advanced views include the posterior edge of the hard palate [Figure 4a] and the posterior part of the soft palate [Figure 4b]. To assess the fetal lip, we obtain the coronal view, allowing us to evaluate the amount and symmetry of the nostrils, as well as the integrity of the upper lip. To visualize the maxillary alveolus, we slide the probe on the axial view from the eye socket toward the fetal chest. The maxillary alveolus presents as a smooth alveolar ridge with tooth buds in morphologically normal cases. To evaluate the

posterior edge of the hard palate, a slight tilt of the transducer toward the fetal chest is required. It is crucial to ensure that the transducer's angle of insonation remains below 30°.[12] While keeping the probe on the view of the posterior edge of the hard palate, a gentle swing toward the fetal chest enables us to capture the posterior part of the soft palate. In this view, we carefully observe the soft palate, and to confirm its integrity, we record a video clip of amniotic fluid passage during fetal swallowing.[11] During the resting phase, the tongue root appears adjacent to the soft palate with similar echogenicity. However, amniotic fluid passes through the oropharynx during swallowing, separating the tongue root and soft palate, which allows us to assess the integrity of the soft palate. The method was described by our team elsewhere.[11]

The described specialized views and techniques contribute to the comprehensive evaluation of the fetal lip and palate, including soft palate, enabling the detection of abnormal structures and the determination of the OFC type.

Examples of Detected Orofacial Clefts

OFCs vary in severity and involvement of associated structures. They can occur in isolation or coexist with other abnormalities. The primary goal of prenatal ultrasound screening is to furnish comprehensive information to the multidisciplinary team regarding the type of cleft, enabling the development of a treatment plan and counseling for parents. The timing of OFC screening is critical for obtaining optimal images of the involved anatomical structures. In cases with a family history of OFCs, first-trimester evaluation of the palate and alveolus might be recommended. However, additional lip imaging is still necessary after 18 weeks of gestation, as ultrasound visualization of fetal lips and the soft palate may be obscured at earlier fetal developmental stages. Given that most fetuses undergo routine second-trimester screening between 20 and 24 weeks of gestation, and visibility of orofacial fetal structures is optimal at this stage, a second-trimester anatomical scan is ideal for OFC screening.

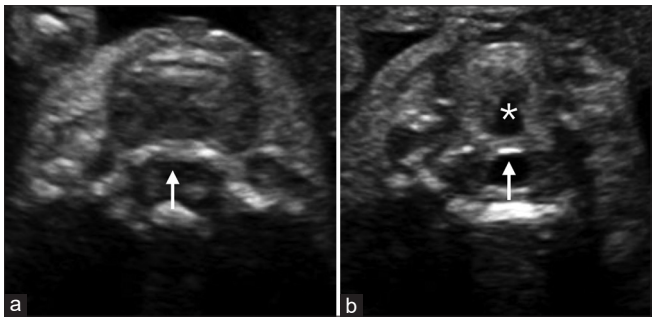

Figure 4: A series of two-dimensional grayscale ultrasound images for advanced orofacial cleft screening. Additional advanced screening views include the posterior edge of the hard palate (arrow, a) and soft palate (arrow, b). The space between the tongue root and soft palate becomes observable during fetal swallowing (asterisk, b). The applied approach for the assessment of the soft palate during fetal swallowing was described in detail elsewhere[11]

During OFC screening, obtaining a coronal view of the nose-mouth area to visualize the lip and an axial view of the alveolus and palate is essential. Variations in laterality – unilateral (u) or bilateral (b) – depend on the presentation of the defect on one or both sides. If a lip defect is observable on the coronal view while other views remain intact, it suggests a type 1 cleft. When a cleft affects both the lip and alveolus, assessing the posterior edge of the hard palate is necessary. An intact hard palate corresponds to type 2, while the entire cleft from the lip to the soft palate is type 3. A disrupted hard palate combines with the cleft soft palate, which can be observed during fetal swallowing. The nostril floor should be examined when the nostrils and alveolus are visible in the same view, specifically on the oblique axial view. If the noncleft side's alveolus protrudes into the cleft side's nasal cavity, indicating a complete form, a disrupted alveolus is evident. Conversely, an intact skin line of the nostril floor signifies an incomplete form [Figure 5].

In type 3u cases, the vomer bone deviates toward the noncleft side, visible in the oral cavity, while the central position of the vomer bone characterizes type 3b cases. The distinction between type 3 and 2 + 4 may warrant discussion [Figure 6]. Notably, type 3u complete form, type 3u incomplete form, and type 2u + 4 exhibit similar features on the lip, alveolus, and soft palate views, though the defect of lip and alveolus can appear wider in type 3u complete form. In type 3u incomplete form, the vomer bone is visible in the oral cavity, with the

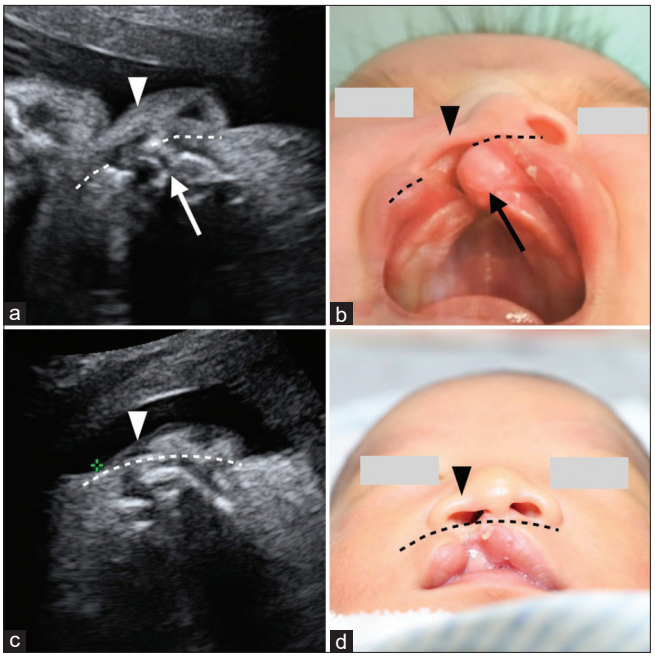

Figure 5: Two-dimensional ultrasound images of lip and upper alveolus on the oblique axial view and corresponding postnatal photographs for complete (a and b) and incomplete (c and d) orofacial clefts. In the complete type (a and b), the affected-side nostril (arrowhead) is collapsed and the noncleft side's alveolus protrudes into the cleft side's nasal cavity (arrow), resulting in the broken nostril floor. In the incomplete type (c and d), the affected-side nostril is smaller (arrowhead) with intact nostril floor (dashed line)

intact nostril floor on the oblique axial view. In type 2u + 4, the vomer bone's visibility is limited, and the margin of the secondary palate cleft can be ascertained.

Fetal swallowing movement should be assessed when the posterior edge of the hard palate appears disrupted while the lip and alveolus are intact. If a cleft is observed during fetal swallowing, it corresponds to a type 4 cleft. In the case of a type 5 cleft, intact mucous membrane and loose soft tissue density of the soft palate are observed during fetal swallowing.[11] The examples for all discussed OFC types are presented in Figure 7, including prenatal ultrasound findings and postnatal photographs.

Conclusion

In conclusion, universal OFC screening is necessary for establishing multidisciplinary management to improve continuity of care and postnatal outcomes. The presented step-wise approach might serve as a basis for developing local guidelines.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Acknowledgments

We would like to thank Ms. Pi-Yuan Sun for medical illustration.

Financial support and sponsorship

The study was funded by the Taiwan Institute of Fetal Medicine (Taiwan Registered Nonprofit Organization 1080280906).

Conflicts of interest

Dr. Tung-Yao Chang, an editor at *Journal of Medical Ultrasound*, had no role in the peer review process of or

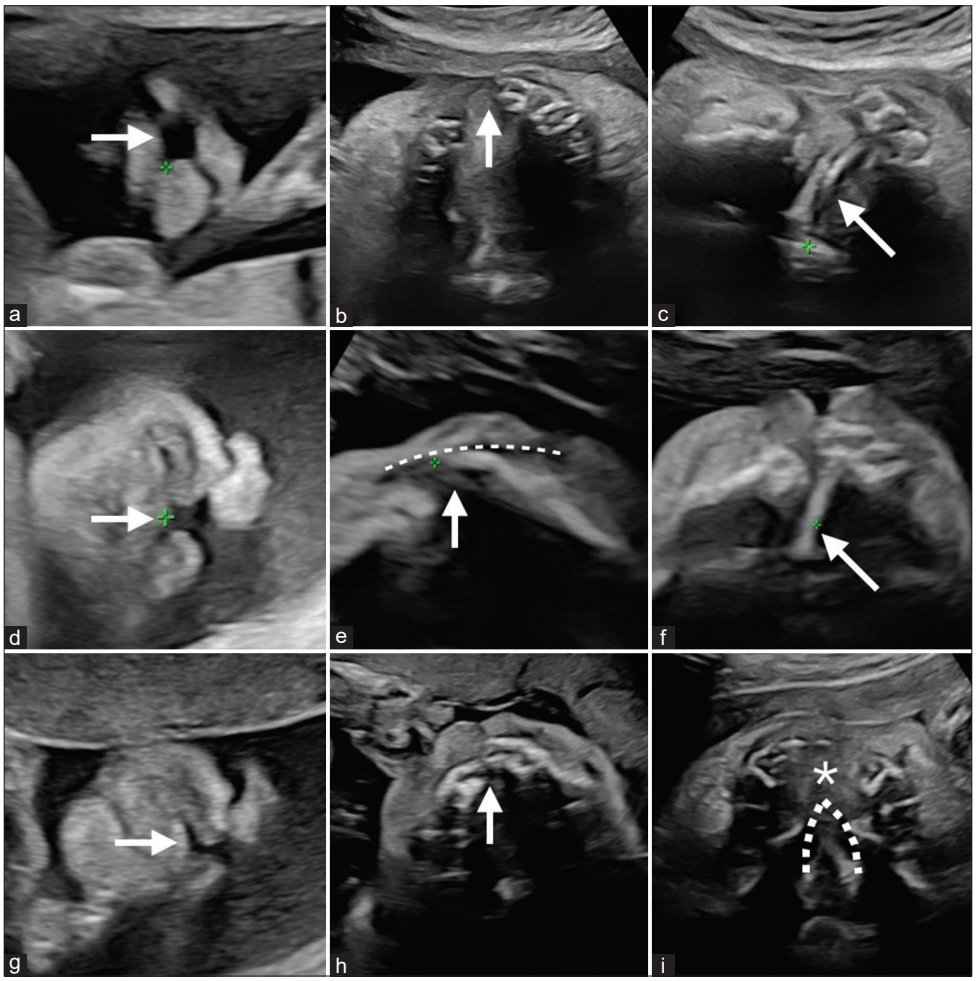

Figure 6: Two-dimensional ultrasound images of the lip, upper alveolus, and the posterior edge of the hard palate on the axial, and oblique axial views in a fetus at 31 weeks of gestation with complete type 3 (a-c); in a fetus at 27 weeks with incomplete type 3 (d-f); and in a fetus at 31 weeks with combined type 2 + 4 (g-i). There are certain similarities in the cleft lip (respectively for a, d, and g, arrow). However, the defect of the alveolus appears wider in complete type 3 (b). The intact nostril floor (e, dashed line) can be observed on the oblique axial view in incomplete type 3. The vomer bone deviation could be observed for incomplete and complete type 3 (c and f, arrow). In type 2 + 4, the vomer bone's visibility is limited, and the margin of the secondary palate cleft (i, dashed line) and the remaining palate (i, asterisk) can be ascertained

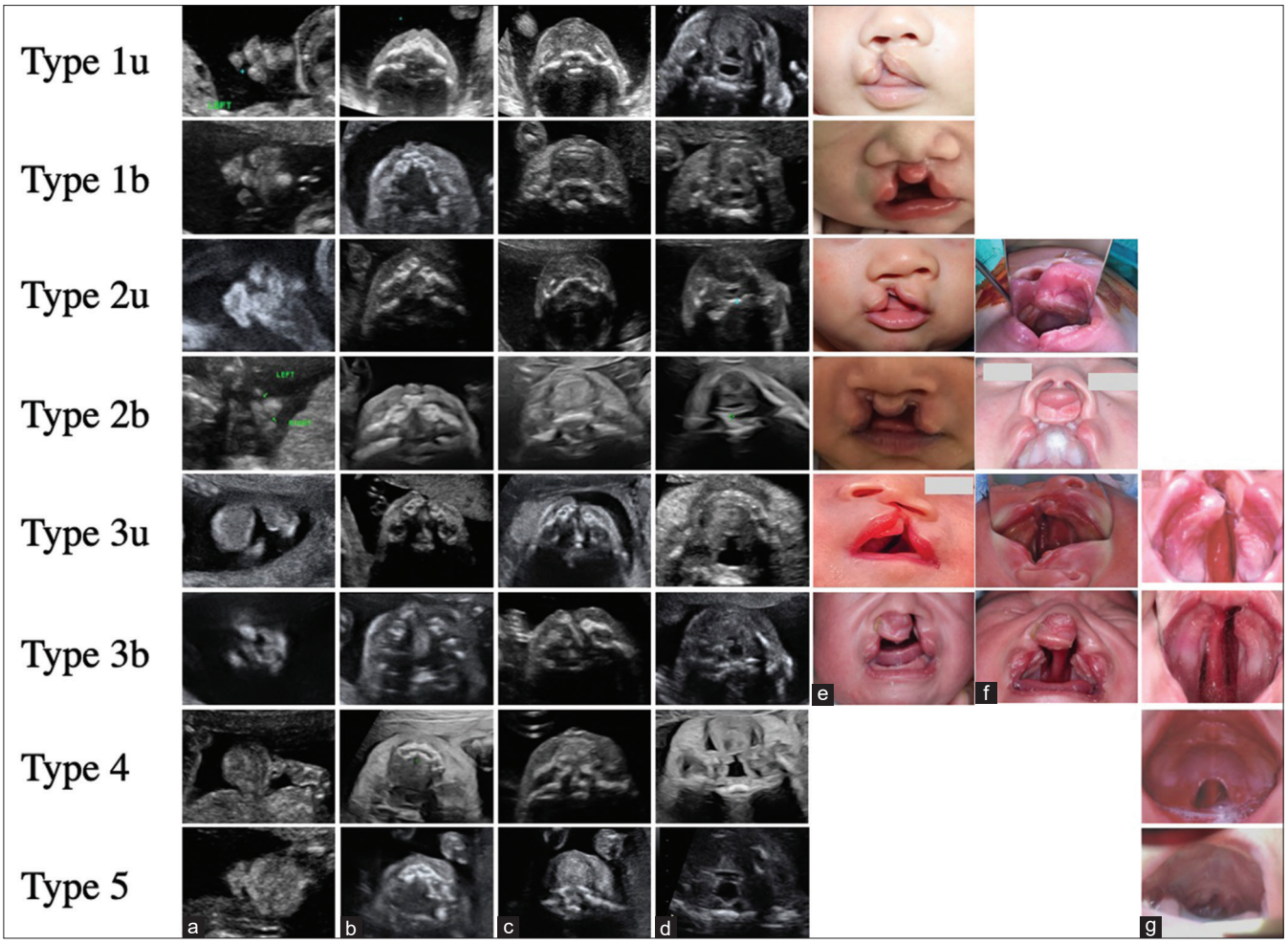

Figure 7: Prenatal 2D ultrasound images of the orofacial clefts according to Maarse *et al*. classification in basic axial view, coronal view, sagittal view, and advanced oblique axial view with corresponding postnatal pictures. Prenatal 2D ultrasound images: (a)-lip; (b)-maxillary alveolus; (c)-posterior edge of hard palate; (d)-posterior part of soft palate. Type 1: A (+) B (−) C (−) D (−) cleft lip with the intact alveolus, hard palate and soft palate. Type 2: A (+) B (+) C (−) D (−) cleft lip and alveolus with the intact hard palate and soft palate. Type 3: A (+) B (+) C (+) D (+) cleft lip, alveolus, and palate. Type 4: A (−) B (−) C (+) D(+), intact lip and alveolus with the cleft hard palate and soft palate during fetal swallowing. Type 5: A (−) B (−) C (+) D (+/−), intact lip, alveolus with disrupted posterior edge of the hard palate and intact mucous membrane of soft palate during fetal swallowing. The corresponding postnatal photographs: (e)-lip; (f)-alveolus; (g)-palate

decision to publish this article. The other author declared no conflict of interest in writing this paper.

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