



Case Report

Hysteroscopic fetoscopy: A role as virtuopsy for parents who refuse full autopsy? A case of facial clefting, proboscis, and limb deformities



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ABSTRACT

Objective: To describe the value of hysteroscopic fetoscopy (*virtuopsy*) at the time of uterine suction in a case of early diagnosis of congenital anomalies in parents refusing conventional full autopsy examination.

Case report: First trimester ultrasound diagnosis of proboscis, median cleft lip and palate and limb deformities. Chorionic villus sampling demonstrated normal karyotype. Parents refused medical induction of termination of pregnancy with subsequent conventional autopsy. At this stage, hysteroscopic fetoscopy was consented and carried out under local anesthesia prior to uterine evacuation.

Conclusion: Hysteroscopic fetoscopy (*virtuopsy*) proved to be a valuable complementary diagnostic investigation and enhanced the parental bonding process concerning the fetal phenotype. Notwithstanding, the woman declared an acceptable compliance during the procedure. In selected cases, *virtuopsy* may be a valid option in confirming early prenatal ultrasound diagnosis in parents refusing conventional autopsy or when full postmortem examination may not be clinically indicated or warranted.

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Introduction

Proboscis may be attributed to a lesion occurring at embryological stage 13, which in the Carnegie staging system corresponds to an embryo with maximum length of 4–6 mm at approximately 32 days postfertilization. Convergence of the nasal disks indicates a head with a narrow median region, a nasal septum that fails to develop, and a single nasal cavity with a single nostril [1].

The lip usually closes by Week 4 of gestation and the palate by Week 12; specifically, the primary palate fuses between 4 weeks of gestation and 6 weeks of gestation, whereas the secondary palate fuses between Weeks 8 and 12. The unilateral cleft lip results from the failure of the maxillary process to close with the medial nasal prominence, and the clefting of the secondary palate is due to the failure of the palatine process to elevate or grow. Cleft palate always starts at the uvula (*uvula bifida* as the mildest form) and proceeds anteriorly along the

midline to affect either the soft palate only or both the soft and hard palates, whereas cleft lip and palate always starts at the lip and proceeds dorsally (alveolus, hard palate, and soft palate) [2].

Case Report

A 28-year-old primigravida with a body mass index (BMI) of 40 kg/m² underwent first-trimester screening for Down syndrome at 12 weeks and 1 day of gestation. The ultrasound examination was performed using a Voluson E8 ultrasound apparatus (GE, Healthcare Medical System, Milwaukee, WI, USA) with a 4–8-MHz RAB4D multifrequency probe. Two-dimensional (2D) trans-abdominal ultrasound performed on the sagittal and coronal planes of the fetal face revealed an abnormal profile. This was followed by three-dimensional (3D) ultrasound in the multiplanar mode with volume rendering and tomographic ultrasound imaging (TUI), which was more effective than 2D ultrasound and led to a diagnosis of orofacial clefting (cleft lip *plus* cleft palate) associated with proboscis (Figure 1).

TUI was performed using thin (1-mm) slices with an interval of –3 mm to 3 mm. The facial cleft was displayed on reference line 0 (green reference dot), and the proboscis was shown on lines +2

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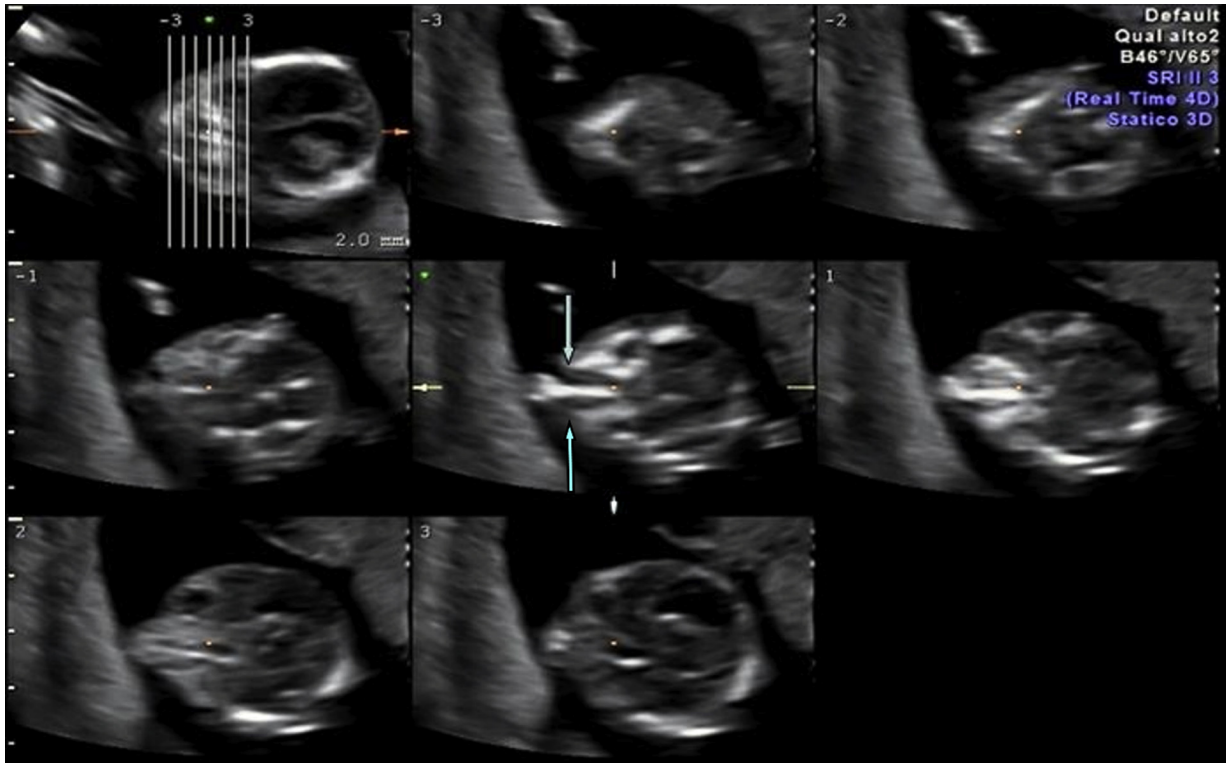


Figure 1. Tomographic ultrasound imaging of the fetal face (comprising the portion from the fetal orbits to the fetal chin), performed using thin (1-mm) slices, revealed median clefting (cleft lip plus cleft palate; marked by arrows).

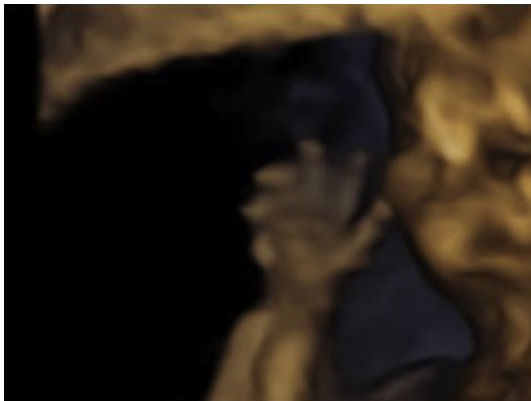


Figure 2. Three-dimensional ultrasound with surface rendering mode showing the proboscis and polydactyly of the right hand.

and +3. Limb deformities involving the upper limbs were detected; however, due to the high maternal BMI of 40 kg/m², precise resolution of this area was not possible. Polydactyly of the right hand and syndactyly of the left hand were identified on ultrasound (Figure 2).

Genetic counseling was offered, and chorionic villus sampling performed at 12 weeks and 2 days of gestation revealed a 46,XY fetus. Because of the major fetal congenital abnormalities, the parents requested termination of pregnancy, but refused administration of vaginal prostaglandin E and subsequent full necropsy examination; however, the mother consented to undergo hysteroscopic embryoscopy (*virtuopsy*) under local anesthesia prior to uterine evacuation.

The woman was placed in the dorsal lithotomy position, and the cervix and the vagina were cleansed with Betadine. One ampoule of

lidocaine was injected laterally into the cervix. After dilation of the cervical canal until Hegar 5 mm, a rigid BETTOCCHI hysteroscope (Karl Storz, Germany) with a 12° angle view and both biopsy and irrigation channels was inserted transcervically into the uterine cavity. Continuous normal saline flow was maintained throughout the procedure (40–160 mmHg pressure) to help distend and clean the uterine cavity. The chorion and the amnion were opened using microscissors to obtain a detailed view of the embryo [3]. All procedures were displayed on a television monitor to enhance the mother's understanding (Figures 3A, B, and 4) and were recorded for offline viewing and analysis.

Surgical termination of pregnancy was accomplished by vacuum curettage of the uterus under general anesthesia.

Discussion

Although 3D ultrasound with volume rendering and TUI-enhanced 2D ultrasound detected lesions involving either the soft tissue (cleft lip and proboscis) or the hard palate (cleft palate), hysteroscopic fetoscopy (*virtuopsy*) provided a direct, extremely clear view of fetal abnormalities and allowed digital recording for subsequent offline analysis.

Considering that the parents refused a conventional full autopsy, *virtuopsy* had a dramatic clinical impact in guiding and aiding postprocedure genetic counseling.

In a previous study by Yin et al [4], which included 12 pregnant women scheduled for legal termination of pregnancy at 6–12 weeks' gestation, successful embryofetoscopies with clear visualization of the embryo or fetus were achieved in 50% of cases. Philipp and Kalousek [5] reported general embryonic maldevelopment in 31% of the cases by transcervical embryoscopy performed prior to evacuation in 154 cases of missed abortion. In that study, chromosomal abnormalities were found

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