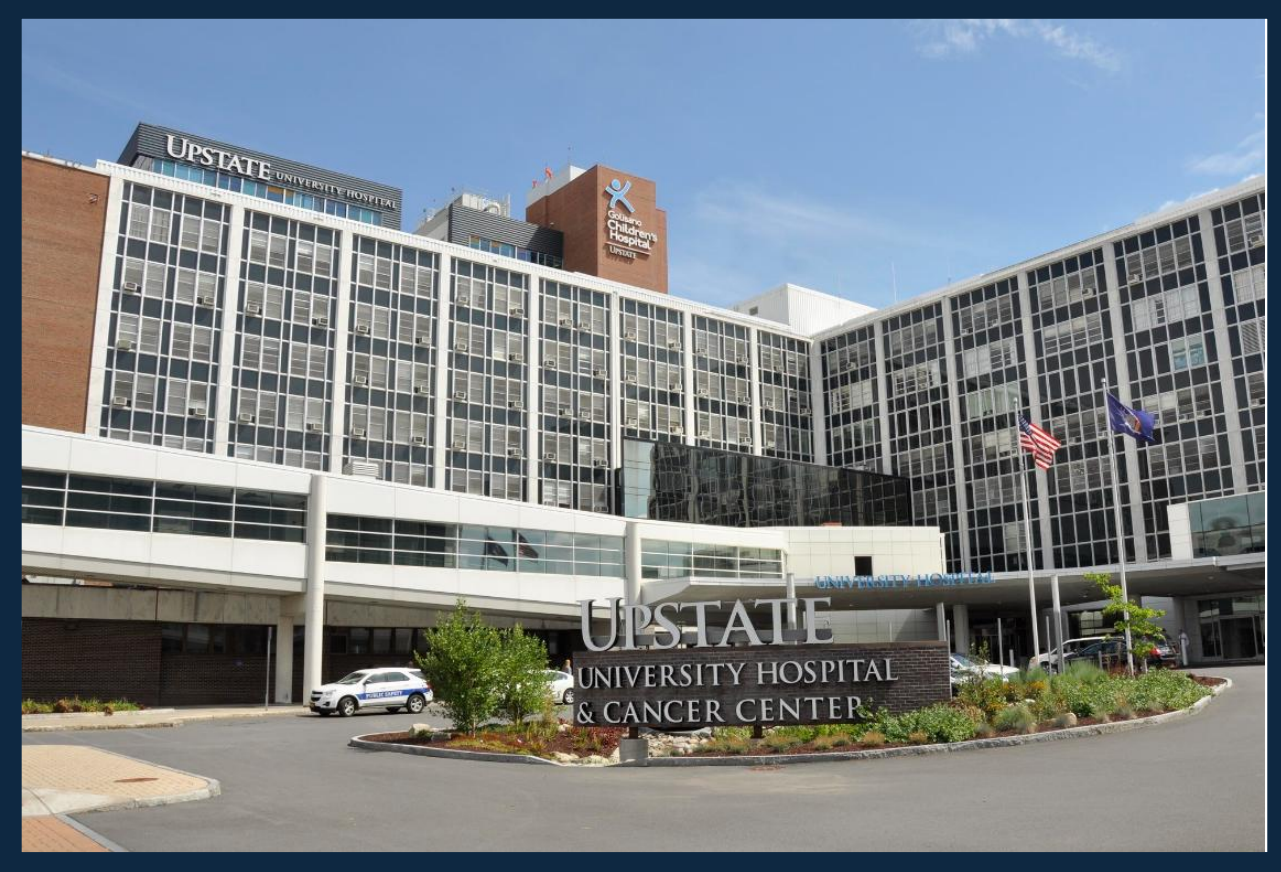




EP05.07 Advanced three-dimensional imaging of bilateral persistent fetal vasculature in association with a pathologic variant in the NDP gene



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Introduction:

The presence of the embryonic hyaloid vascular network within the developing globe is a normal part of human development up to 30 weeks of gestation at which time it undergoes spontaneous regression in normal fetuses. The persistence of these vessels in the latter part of the third trimester, particularly the hyaloid artery, is considered abnormal and is associated with a variety of congenital ocular defects which may lead to serious vision loss or blindness¹. Ultrasound diagnosis of persistent (PFV) is achieved by demonstrating blood flow with color Doppler in the hyaloid artery within the fibrovascular stalk that extends from the optic disc to the posterior surface of the lens. Advanced three-dimensional imaging can assist in the prenatal diagnosis of this condition.

Case:

At 37 weeks of gestation, a complex, disorganized accumulation of structures were noted within the normally anechoic globes. Bilateral linear striations or “septations” were demonstrated within the vitreous cavity representing retinal detachment and a fibrovascular stalk was noted extending from the posterior surface of the lens to the posterior aspect of the globe. SlowflowHD color Doppler identified blood flow within the persistent hyaloid artery (Figure 1). Three-dimensional sonography was used to further evaluate the ocular anomalies (Figure 2). A modified reverse face technique was performed and reconstructed images were acquired with traditional three-dimensional surface rendering, Hdlive, and Hdlive silhouette manipulation (Figure 3). These techniques allowed for the unique visualization of the ocular contents from the posterior aspect of the fetal face. Conical fibrous stalks were noted to extend from the posterior surface of the lens to the posterior aspect of the globe. Further manipulation of the thick slice reverse face image of utilized rotational and silhouette adjustments. This technique allowed for a more detailed representation of the fibrovascular stalks and lenses (Figure 4). Based on the three-dimensional sonographic findings, the prenatal diagnosis of bilateral persistent fetal vasculature was suspected. Postnatal pediatric ophthalmic evaluation confirmed the prenatal diagnosis.

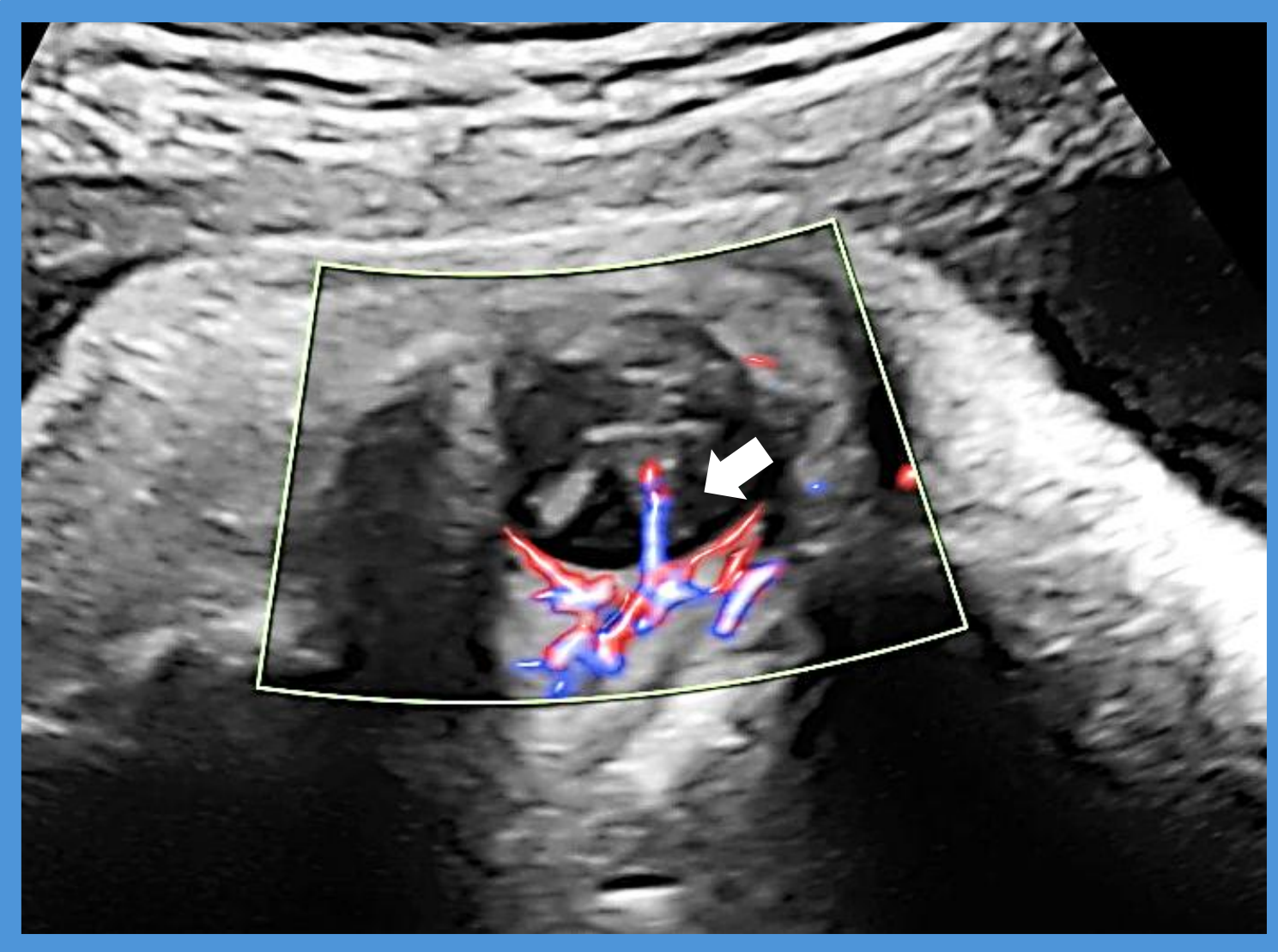


Figure 1: SlowflowHD color Doppler demonstrates blood flow within the hyaloid artery (arrow).

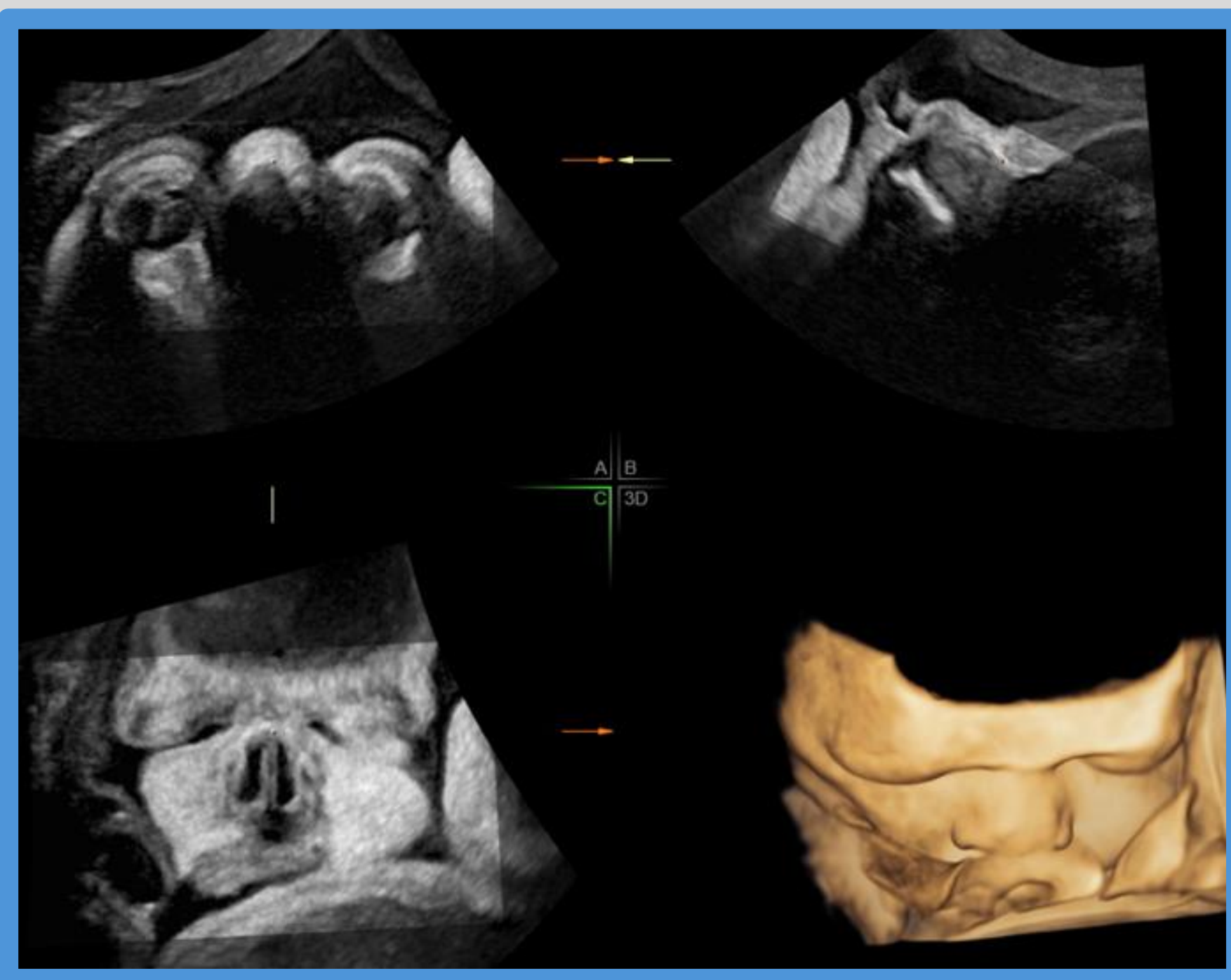


Figure 2: Initial three-dimensional imaging demonstrating PFV within the globes in the image of acquisition but the defect cannot be appreciated in the surface rendered image.

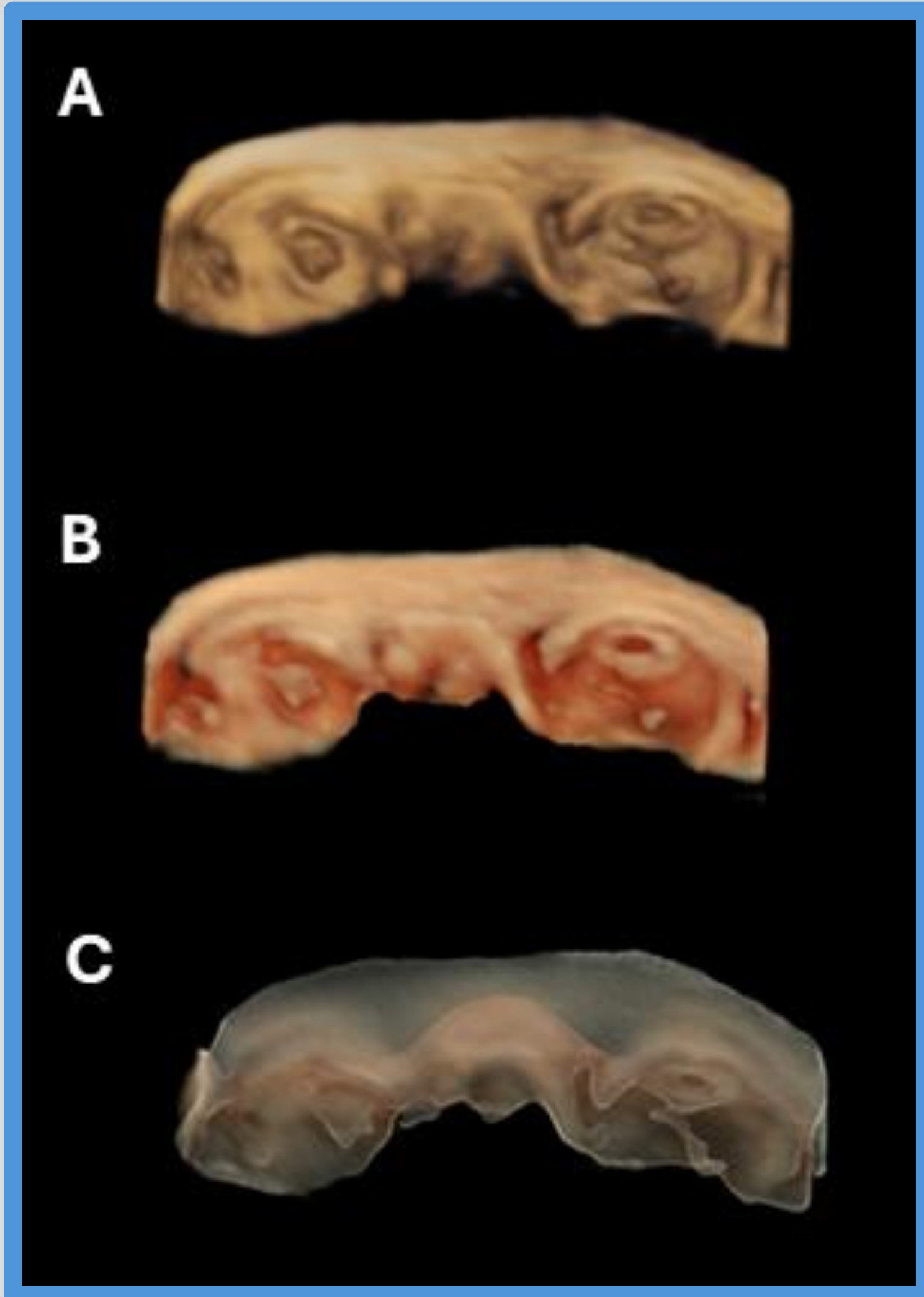


Figure 3: Thick slice modified reverse face technique. (A) Traditional three-dimensional surface rendering. (B) Hdlive application. (C) Hdlive image with silhouette enhancement.



Figure 4: (A)) A thick slice modified reverse face technique with Hdlive silhouette manipulation demonstrates the bilateral fibrovascular stalks (white arrows). (B) The same image as A with a rotational adjustment to optimize visualization of the the right lens (white arrowhead).

Genetics:

Whole genome analysis of the infant’s DNA revealed a hemizygous mutation, c.236_239dup, of the NDP gene (NM_000266.4). This variant was also was found to be heterozygous in the mother and was not detected in the father, consistent with an X-linked recessive inheritance of the NDP-related disorders. Pathogenic variants in this gene have been reported to cause X-linked NDP-related retinopathies. The spectrum of NDP-related retinopathies appears to have considerable overlap, ranging from Norrie disease, NDP-related familial exudative vitreoretinopathy, NDP-related Coats disease, NDP-related advanced retinopathy of prematurity, and as in this case, NDP-related persistent fetal vasculature (Variantyx).

Conclusion:

The intraocular fetal vascular system is critical for the development of the lens, vitreous, and retina. Partial or incomplete failure of its regression can produce different clinical manifestations of persistent fetal vasculature which can be diagnosed by prenatal ultrasound in the late third trimester². This case presents the prenatal diagnosis of bilateral persistent fetal vasculature at 37 weeks of gestation. Subsequent whole genome analysis of the newborn detected a pathologic variant of the NDP gene with X-linked recessive inheritance which has a known association with ocular/retinal anomalies. Advanced three-dimensional imaging techniques provided a unique opportunity to depict this anomaly in various reconstructed formats.

References:
1. Hata T, Koyanagi A, Yamanishi T, Bouno S, Takayoshi R, Miyake T. Natural course of fetal hyaloid artery. J Ultrasound Med. 2022;41:2259-2267.
2. Chen C, Xiao H, Ding X. Persistent fetal vasculature. Asia Pac J Ophthalmol. 2019;8:86-95.