

EP05.56 Prenatal ultrasound diagnosis and clinical analysis of fetal ocular abnormalities



A Single-Center Retrospective Cohort of 83 Cases (2014–2025)

Xiaotong Cai#, Xi Chen#, Weizeng Zheng, Shimin Tu, Kai Yan, Weili Zhao, Yuhan Lou, Xiaojing Wu, Liqun Sun* | Women’s Hospital, Zhejiang University, Hangzhou, China

01. RESEARCH OBJECTIVES

- Characterize prenatal imaging features and genetic profiles of four severe fetal ocular malformations.
- Establish a stratified diagnostic workflow to improve prenatal counseling accuracy.

02. STUDY DESIGN & METHODS

- Retrospective analysis of 83 fetuses with sonographically suspected ocular anomalies (2014–2025).
- Comprehensive data: US, MRI, karyotyping, CMA, trio-WES, and postnatal follow-up.

03. KEY LIMITATIONS

- Single-center design may introduce selection bias.
- High rate of pregnancy termination limits long-term neurodevelopmental follow-up data.

04. POPULATION DISTRIBUTION (n=83)

Microphthalmia (54.2%) was the most common, followed by Congenital Cataract (24.1%), Anophthalmia (14.5%), and PFV (7.2%).

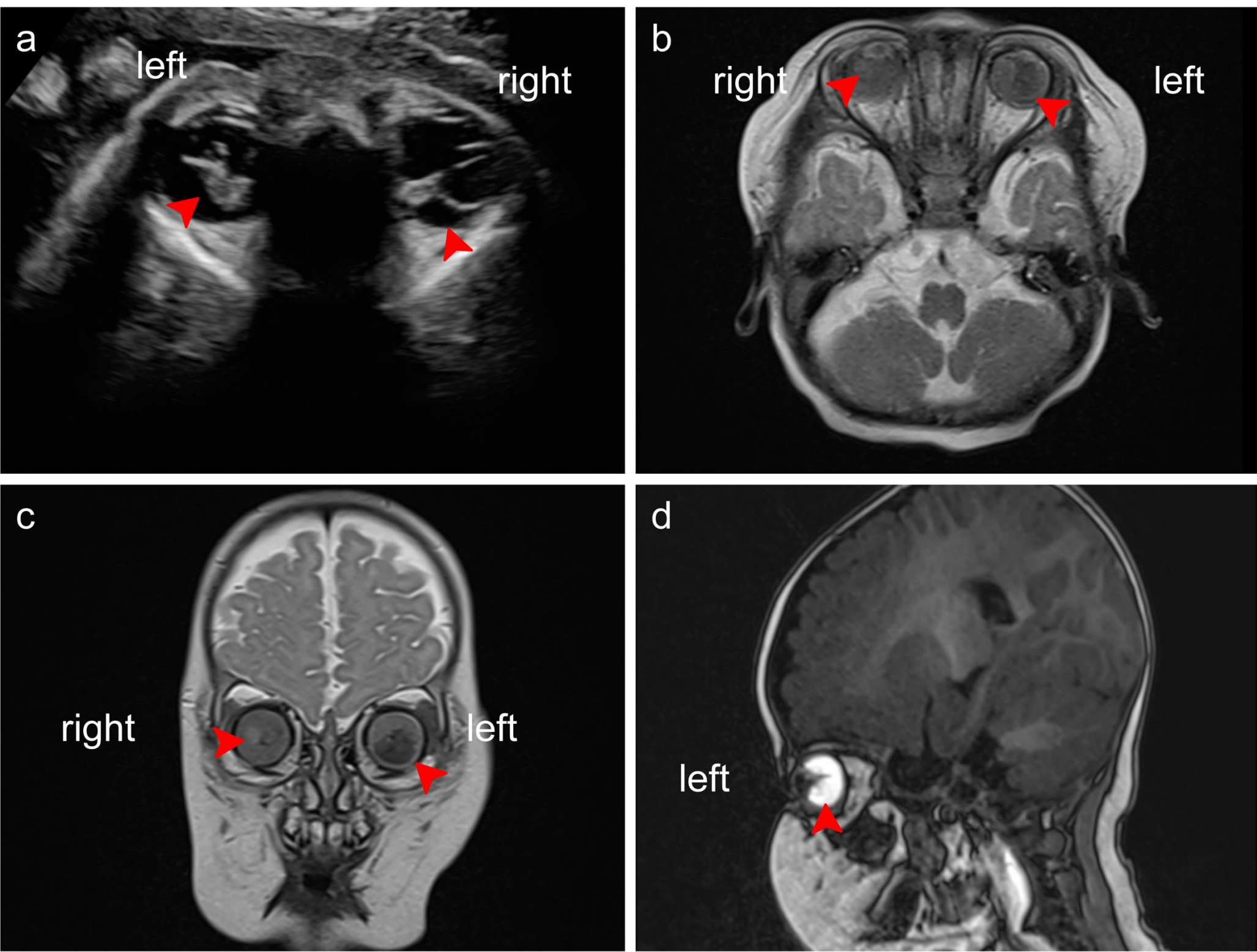


Figure 1. Prenatal ultrasound (a) and fetal MRI (b-d) demonstrating microphthalmia and associated intracranial anomalies. Red arrowheads indicate abnormal ocular structures.

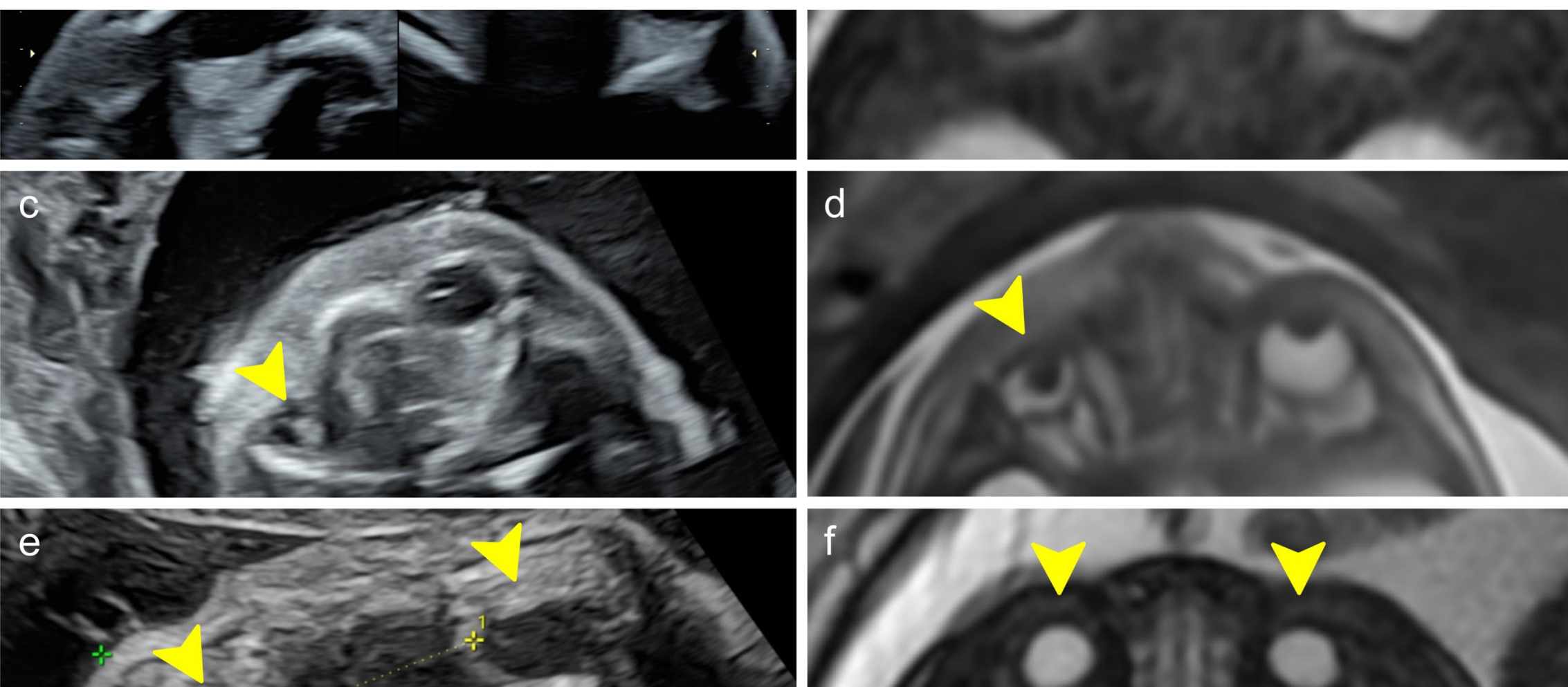


Figure 2. Side-by-side ultrasound (a,c,e) and fetal MRI (b,d,f) correlation of fetal orbital-globe anomalies. Yellow arrowheads mark pathological changes.

05. PERINATAL OUTCOMES

- High TOPFA rates: Anophthalmia (100%), Microphthalmia (95.6%).
- Live-born infants all developed permanent visual impairment.

06. DISCUSSION: KEY INSIGHTS

- **US as Gold Standard:** First-line for detection of all four subtypes.
- **Syndromic Association:** 39.8% had extraocular anomalies (CNS/Cardiac).
- **Genetic Basis:** Variants in the Norrin/Wnt pathway are key drivers.

07. CLINICAL MANAGEMENT

WORKFLOW

SCREENING

Prenatal US

DIAGNOSIS

MRI + WES

COUNSELING

MDT & Prognosis

08. CONCLUSION

Fetal ocular malformations are complex conditions often linked to multisystem anomalies and pathogenic genetic variants. A combined radiological and genomic approach is essential for precise diagnosis and parental counseling.