The Role of Prenatal Imaging in the Identification of Congenital Ocular Anomalies: A Systematic Review

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Background

- Early diagnosis of congenital ocular disease is paramount in optimal postnatal management.

- It is possible to identify ocular anomalies during the prenatal period using imaging tools such as fetal ultrasound (fUS)[1].

- However, standards of care regarding the clinical utility of prenatal imaging in identifying ocular anomalies during the prenatal period are lacking[2].

- This systematic review aims to evaluate the evidence regarding the role of prenatal imaging in identifying ocular and orbital abnormalities.

Methods

- Database searches in Embase, MEDLINE, and Cochrane for keywords and database-specific terminology (e.g., MeSH) were performed without restrictions.

- **Included:** Case reports, case series, and full-length articles describing identification of ocular or orbital anomalies using fUS and/or fetal MRI (fMRI).

- **Excluded:** Review articles, meta-analyses, conference abstracts, and articles without an English translation.

- Two reviewers assessed each reference for eligibility according to predefined criteria. Discrepancies were resolved by a third reviewer.
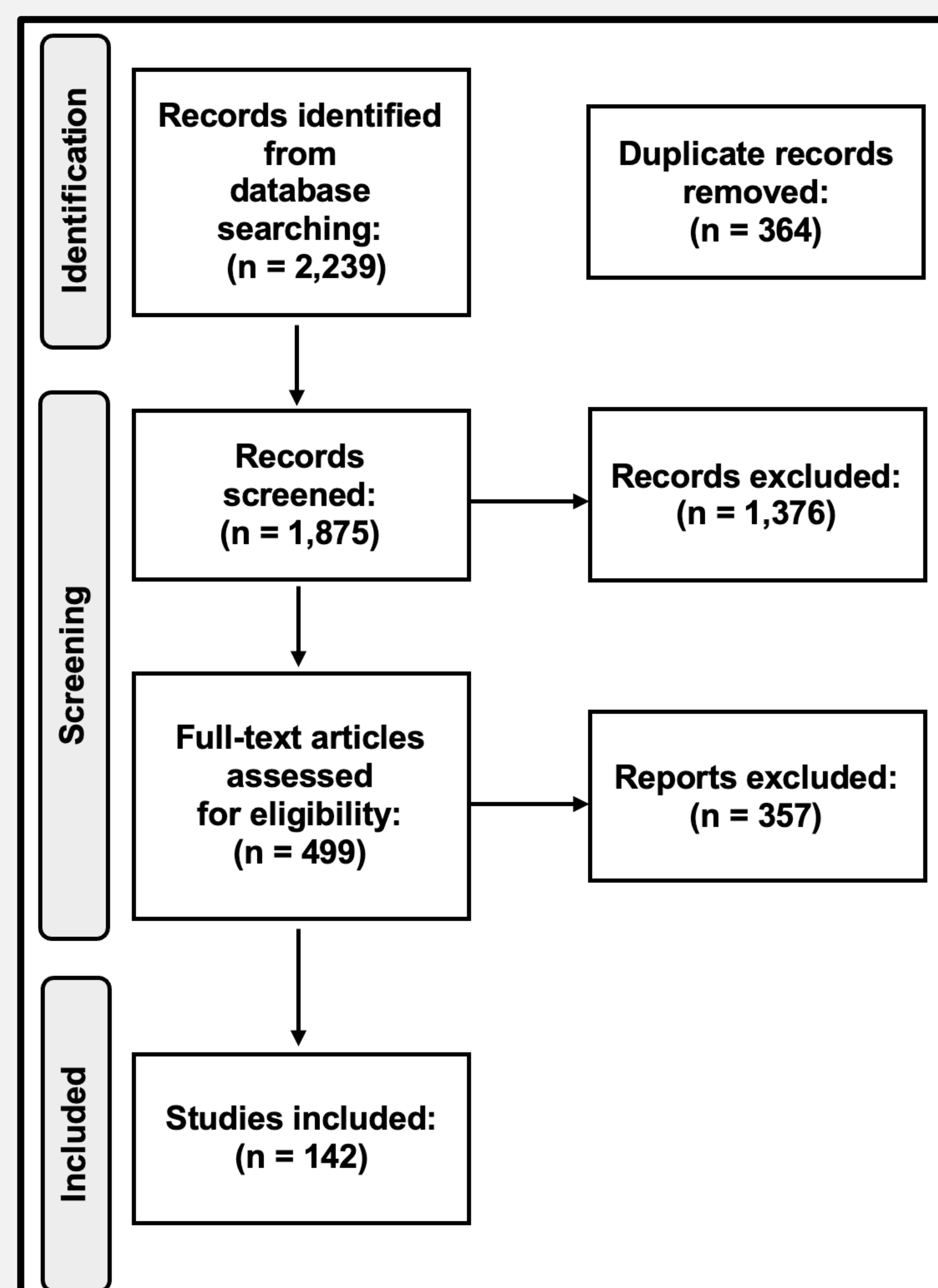
Figure 1. PRISMA diagram depicting search and screening results.

Results

Study Characteristics	Studies (N=142)
Study Type	
Case Reports	110 (77.5%)
Case Series	21 (14.8%)
Retrospective Cohorts	9 (6.3%)
Prospective Cohorts	1 (0.7%)
Continent	
Europe	69 (48.6%)
North America	33 (23.2%)
Asia	32 (22.5%)
Australia	4 (2.8%)
South America	3 (2.1%)
Africa	1 (0.7%)

Subject Characteristics	Subjects (N=314)
Imaging Modality	
fUS	268 (85.4%)
fMRI	17 (5.4%)
fUS + fMRI	29 (9.2%)
Trimester	
1st	3 (1.3%)
2nd	136 (57.4%)
3rd	97 (40.9%)
Gestational Age (weeks)	
Mean (SD)	24 (± 6)
Median (Range)	23 (11 – 38)

Table 1. Overview of all included publications (n = 142).

Ocular Anomalies Identified Prenatally

- Microphthalmia (33.43%)
- Cataract (32.87%)
- Anophthalmia (12.43%)
- Coloboma (8.29%)
- Retinal Dysplasia (3.31%)
- Microphthalmia with Cyst (3.04%)
- PHPV (1.93%)
- Cryptophthalmia (1.66%)
- Glaucoma (1.38%)
- Orbital Tumor/Mass (1.10%)
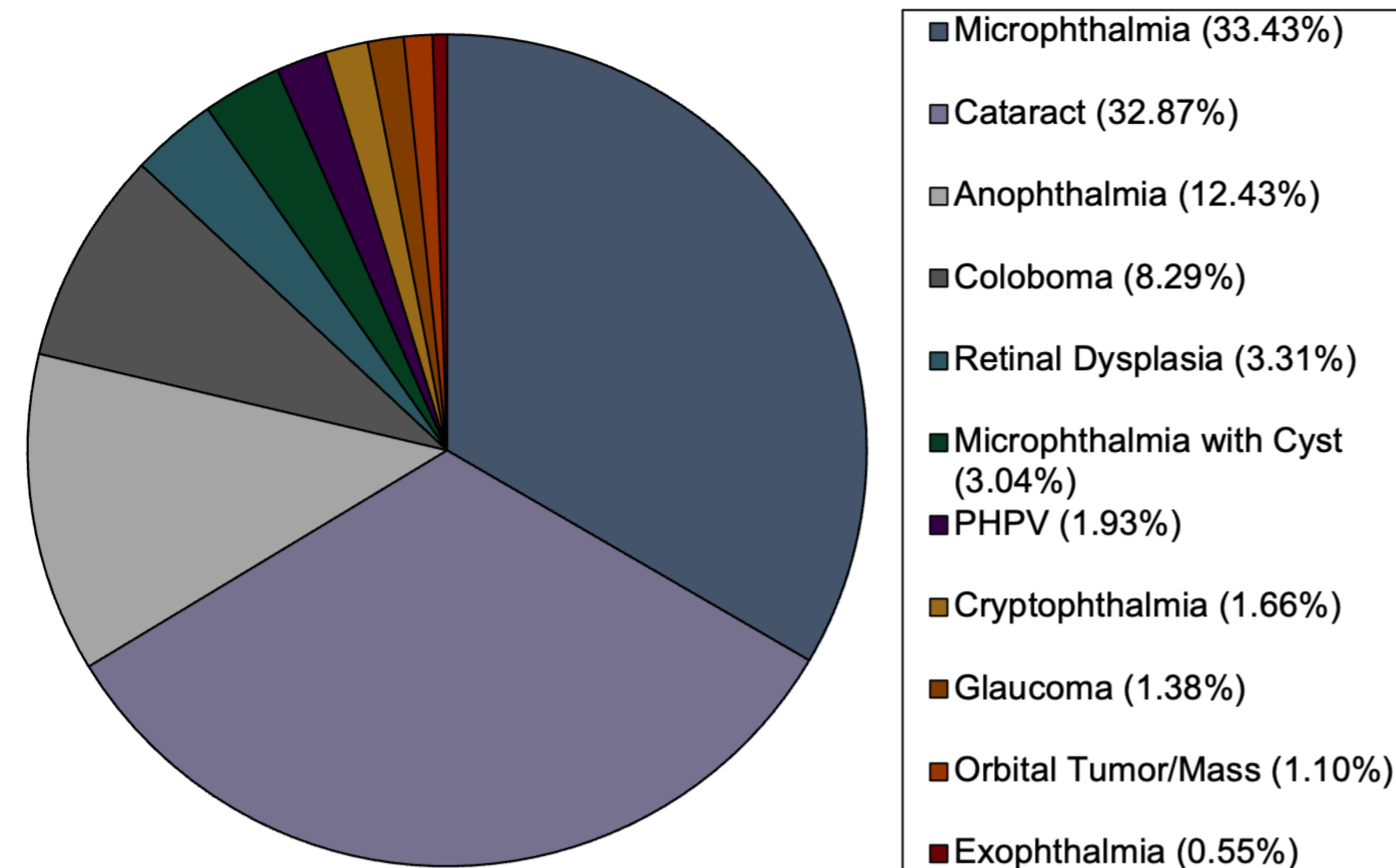- Exophthalmia (0.55%)

Figure 2. Distribution of ocular anomalies identified using prenatal imaging.

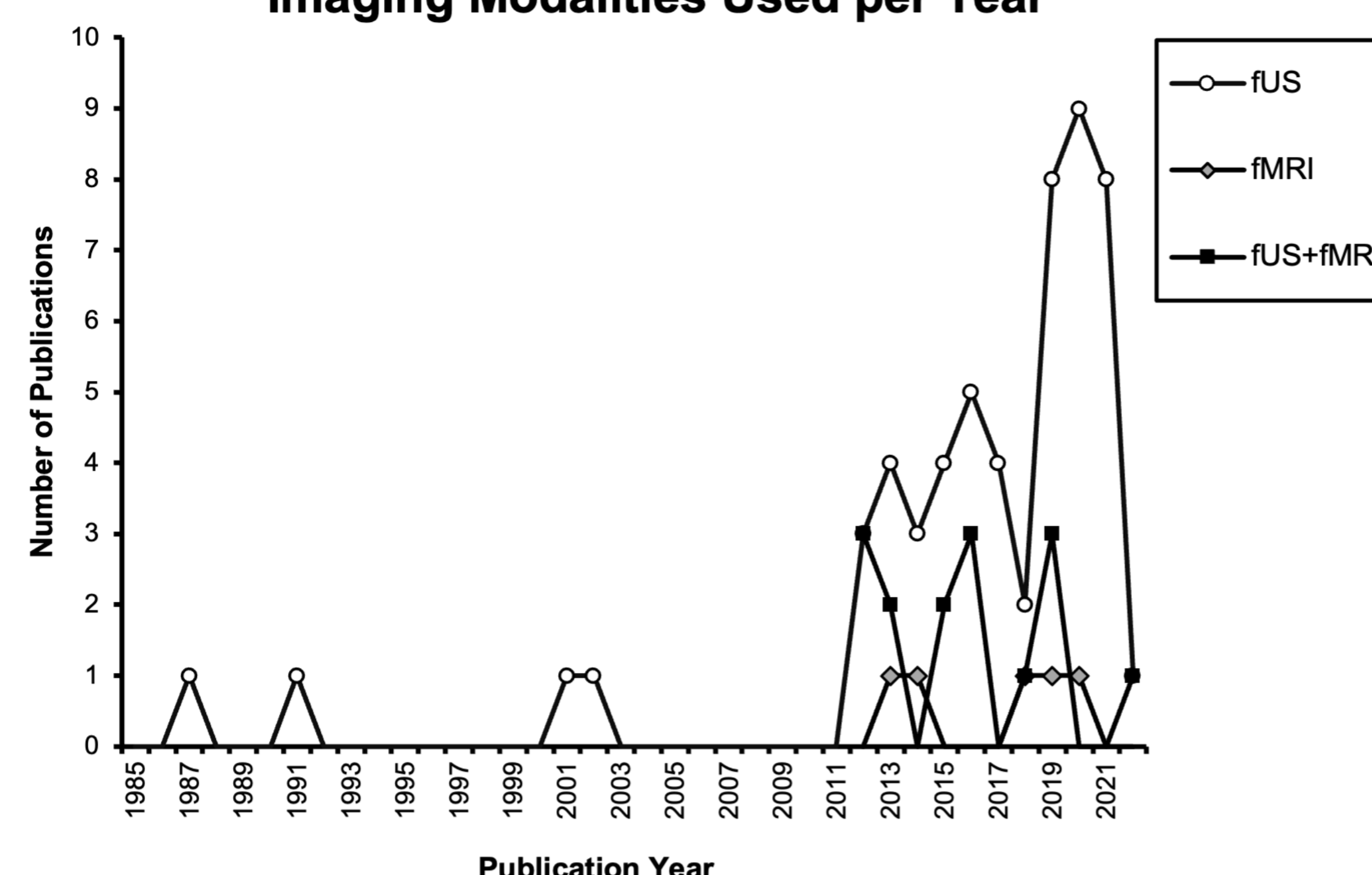**Imaging Modalities Used per Year**

Figure 3. Prenatal imaging modalities used in the identification of ocular and orbital anomalies from 1985-2021.

Results

- The most common diagnoses were cataract, microphthalmia, and anophthalmia.

- Most cases (98.7%) were identified during the 2nd or 3rd trimester.

- fUS alone was used to identify anomalies in most (85%) studies. Multimodal imaging (fMRI/fUS) was used most frequently for posterior segment anomalies.

- Imaging findings informed postnatal surgical and medical treatment in 26 cases (8%).
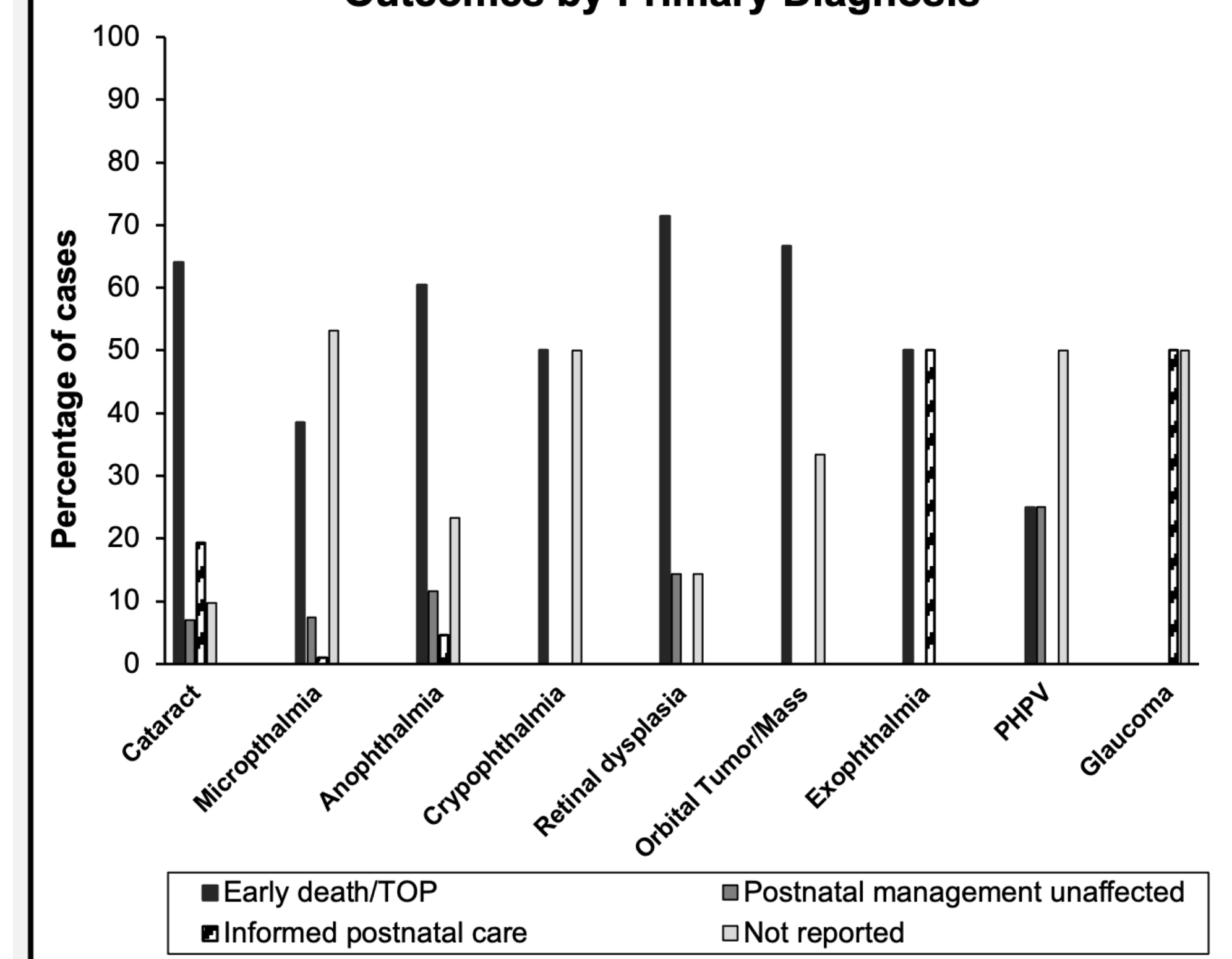
Outcomes by Primary Diagnosis

Figure 4. Postnatal outcomes classified by primary diagnosis.

Conclusions

- A broad spectrum of ocular and orbital anomalies in fetuses have been identified using fetal imaging.

- Prospective, multi-institutional studies are needed to develop clinical standards for prenatal ocular assessment.

- Future work should consider the potential role of maternal-fetal medicine in the identification of ocular and orbital anomalies.

References

1. Qin Y, Zhong X, Wen H, et al. Prenatal Diagnosis of Congenital Cataract: Sonographic Features and Perinatal Outcome in 41 Cases. Pränatale Diagnose des angeborenen Katarakts: Sonografische Merkmale und perinatales Outcome in 41 Fällen. Ultraschall Med. 2022;43(6):e125-e134. doi:10.1055/a-1320-0799

2. Ondeck CL, Pretorius D, McCaulley J, et al. Ultrasonographic prenatal imaging of fetal ocular and orbital abnormalities. Surv Ophthalmol. 2018;63(6):745-753. doi:10.1016/j.survophthal.2018.04.006

Contacts and Acknowledgements