

Fetal MRI in confirmed genetic syndromes: current evidence on its added value beyond ultrasound

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Introduction

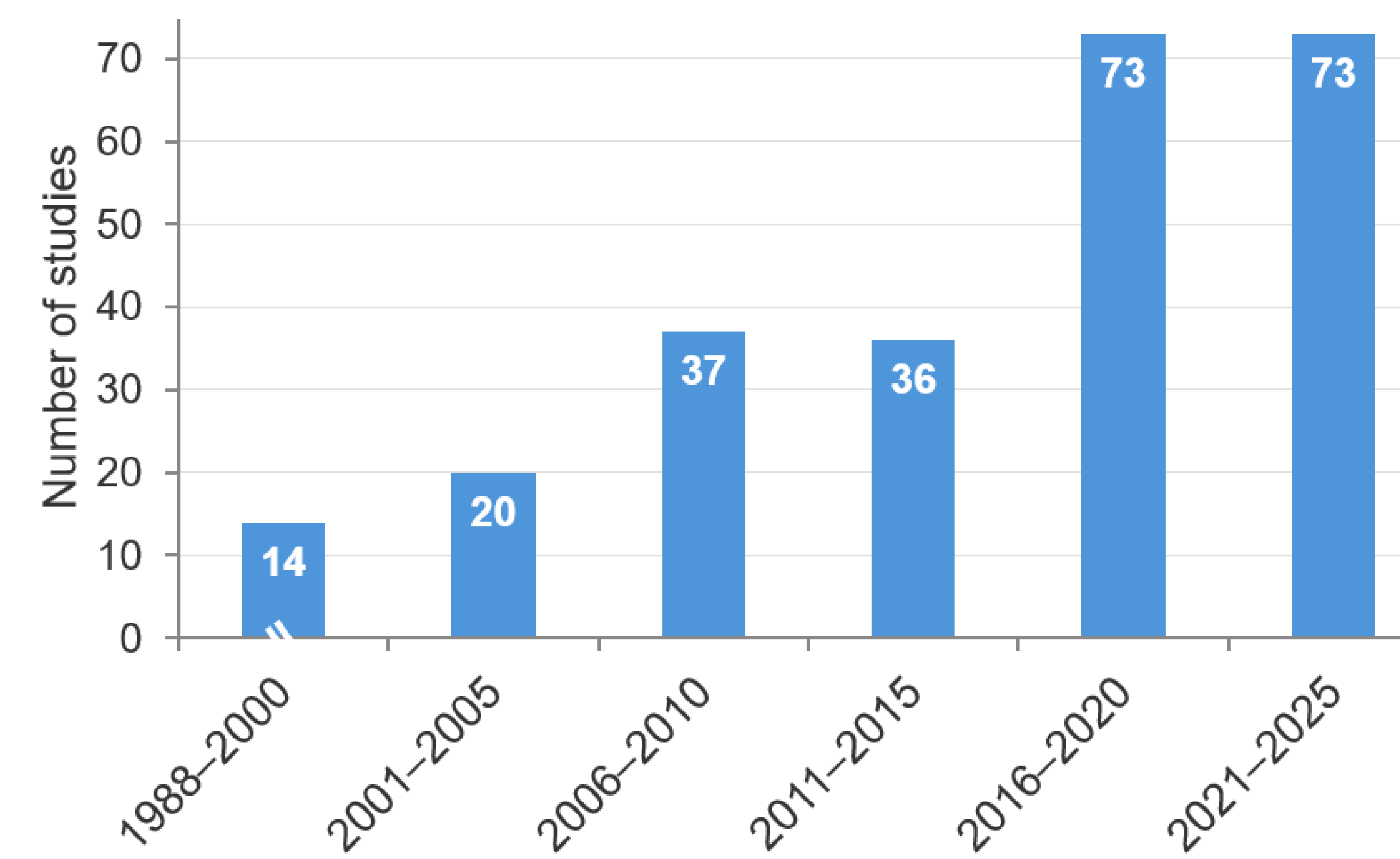
- **Fetal MRI complements ultrasound** in prenatal morphological assessment
- **Accurate prenatal phenotyping** is **critical** for variant interpretation
- However, the **fetal MRI added value** in genotype-phenotype correlation of genetically confirmed syndromes **remains poorly defined**

Methods

- To **evaluate fetal MRI's contribution to genotype-phenotype correlation** in **genetically confirmed syndromes**
- To **quantify its diagnostic added value** compared to ultrasound findings alone

Design

- **Systematic narrative review**, PROSPERO (CRD420261300509)
- Adherence to PRISMA 2020 guidelines
- PubMed/MEDLINE from inception to Nov 2025
- Variables: study type / sample size / phenotypic group / genetic diagnosis / genetic technique / MRI role / MRI contribution



Results

Studies distribution

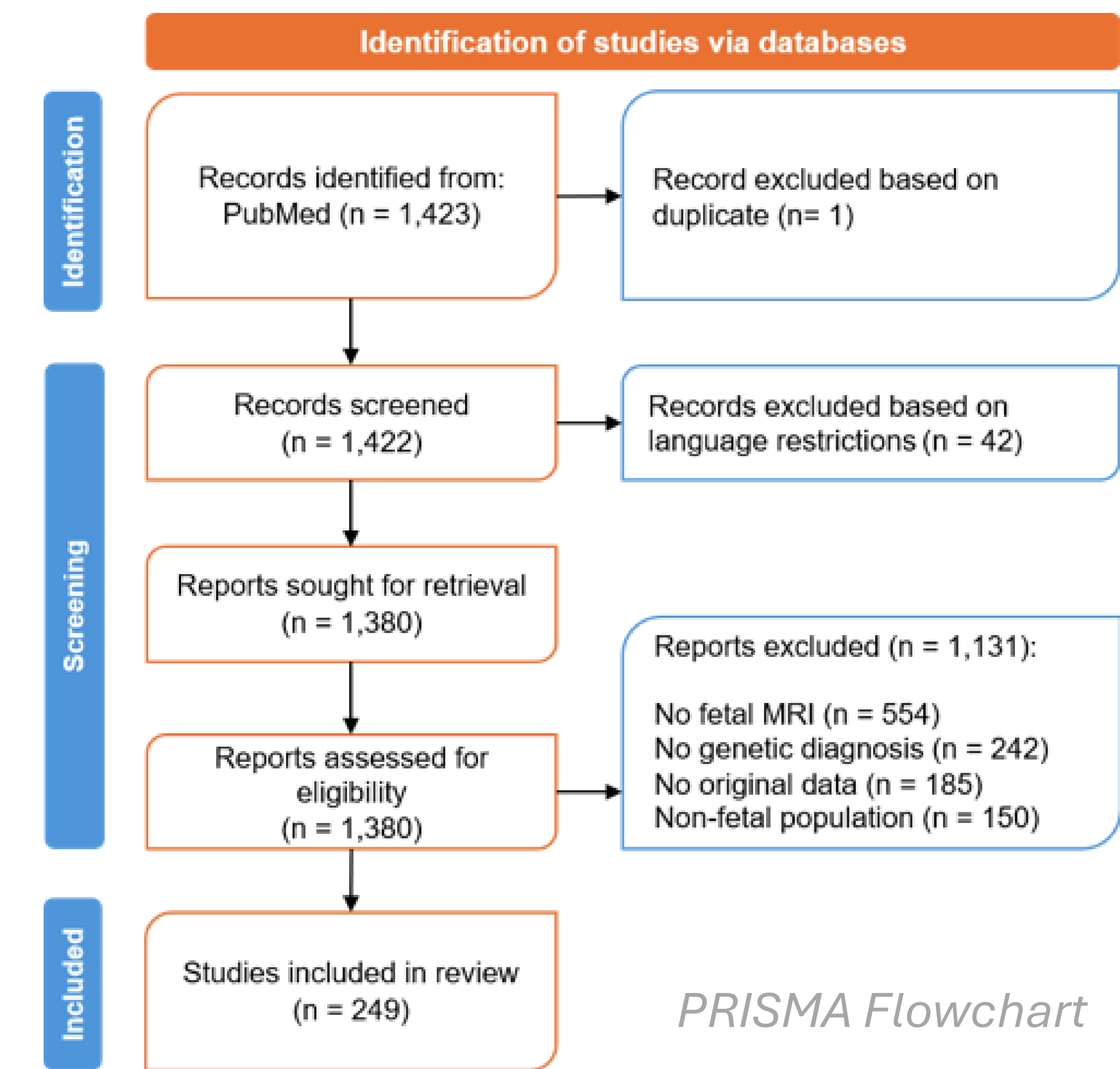
• Case reports : 67% / Cases series : 29 % / Cohorts : 4%
The **predominance of case reports** and **case series** reflects the rarity of genetic syndromes and **inherent difficulty in assembling prospective cohorts**.
While this underscores the **high clinical relevance of individual cases**, it **limits generalizability and strength of evidence**.
It highlights the **need for systematic prospective validation studies** in well-defined genetic entities.

Phenotypic categories

• CNS 45 % / Multisystem 35 % / Other 11 % / Skeletal 6% / Cardiac 3%
CNS predominance aligns with **clinical utility**: fetal MRI adds diagnostic value for CNS anomalies not accessible to ultrasound, justifying **preferential publication and use** in this domain.

Conclusion

This **narrative review of 249 studies**, including **> 1,900 fetuses** with confirmed genetic diagnoses illustrates that :
→ Fetal MRI is **useful across the full spectrum** of genetic syndromes, rather than limited to common entities,
→ Fetal MRI **supported interpretation / oriented testing in ¼ studies**, showing its importance in the genetic workflow,
→ Predominance of **CNS anomalies** (45%) reflects the **greater sensitivity of MRI for brain anomalies** compared to US,
→ Fetal MRI, by refining prenatal phenotypes, is **transitioning from morphological adjunct to dedicated primary tool** for molecular genetic testing, **fundamentally changing how we approach prenatal diagnosis**.



Genetic diagnoses

It reflects breadth of phenotypic involvement :
Tuberous Sclerosis 32 studies / Chromosomal syndromes 29 studies / Skeletal dysplasias 15 studies / **Diverse rare genetic conditions** 103 studies (41%)
Fetal MRI **oriented genetic testing** and/or **supported variant interpretation** in **62 studies** and better described phenotypes in **172 studies**.