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Key message

In low-risk pregnancies with truly isolated ARSA, our cohort showed no additional yield of CMA, while pooled evidence showed a low 2% incremental yield.

Background & objective

Aberrant right subclavian artery (ARSA) is a sonographic marker associated with trisomy 21 and 22q11.2 deletion, but its significance when truly isolated remains uncertain.

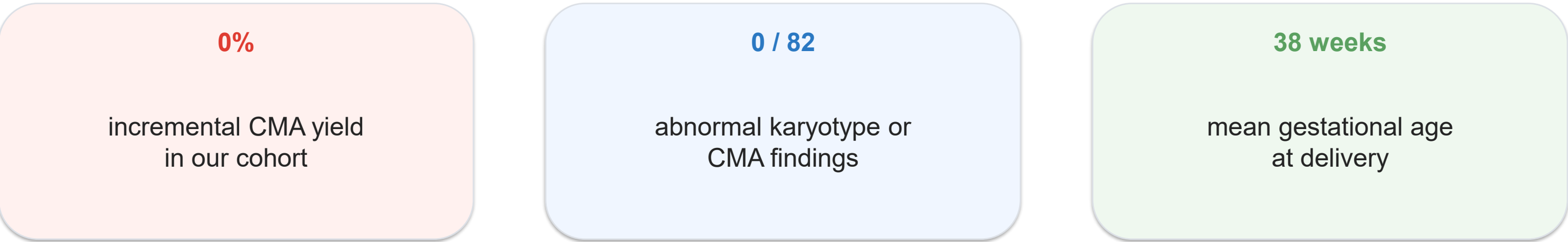
Objective: to quantify the additional diagnostic contribution of chromosomal microarray analysis (CMA) in fetuses with isolated ARSA and normal karyotype using an original cohort and a meta-analysis of the literature.

Cohort study design



Detailed anomaly and cardiac scans were performed in all ARSA cases. Cases with additional major/minor markers or abnormal first-trimester screening results were excluded.

Cohort results

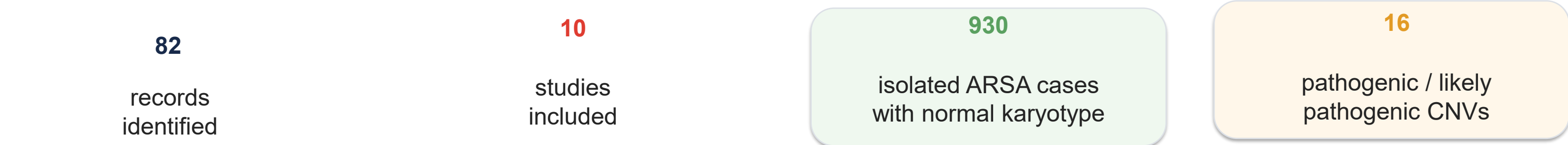


All newborns were healthy, with no obvious congenital anomalies and no reported stridor, dysphagia, intubation, or NICU admission.

Interpretation: in a low-risk setting where ARSA was truly isolated, the cohort provided no evidence of additional diagnostic yield from CMA beyond conventional karyotyping.

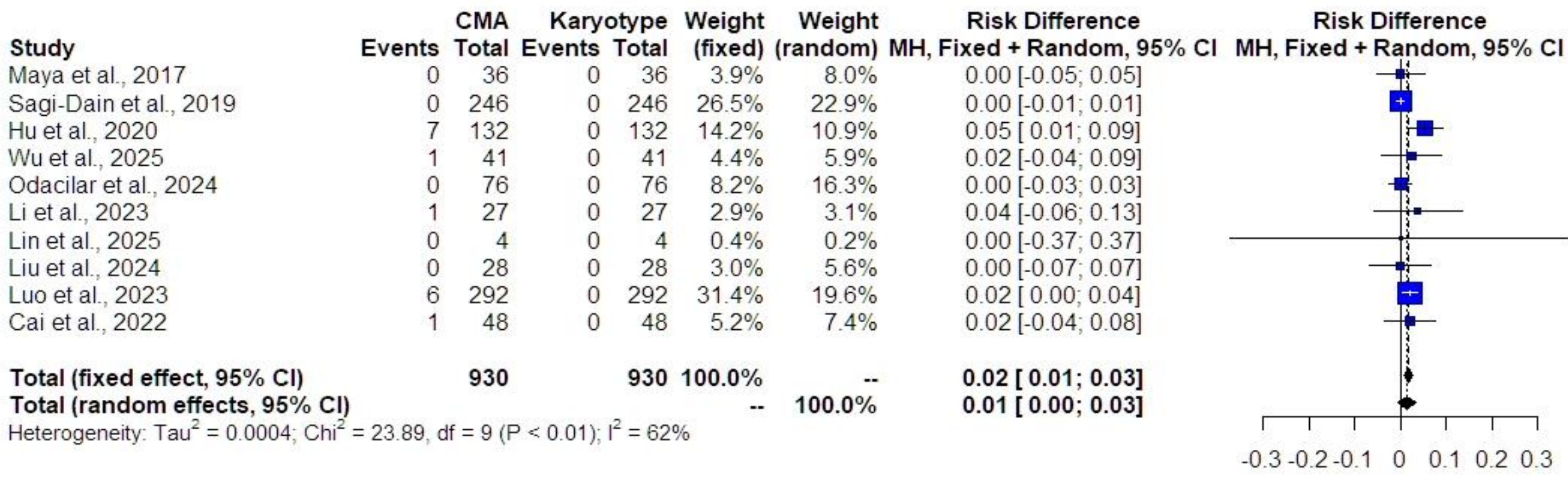
Systematic review & meta-analysis methods

Databases searched: Ovid Medline, Embase, and Cochrane Library through 30 April 2025.
Included studies: observational reports of fetuses with isolated ARSA and normal karyotype undergoing CMA.
Excluded: non-isolated ARSA, case reports/small series, and CNVs ≥10 Mb considered detectable by karyotype.
Outcome: incremental yield (risk difference) of pathogenic / likely pathogenic CNVs detected by CMA beyond conventional karyotyping.



Meta-analysis results

Pooled incremental diagnostic yield of CMA over conventional karyotype in fetuses with isolated ARSA

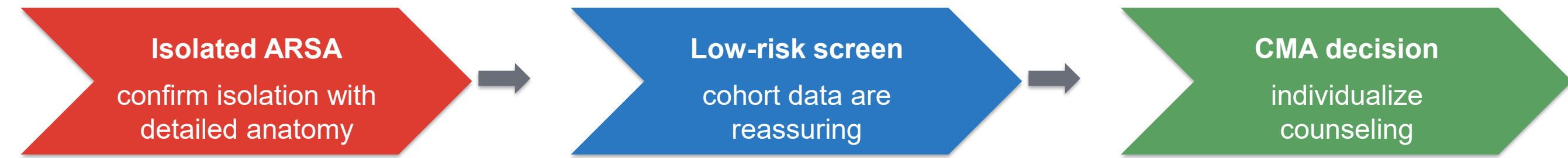


The pooled literature suggests a low but non-zero incremental yield. This estimate should be interpreted cautiously, as some pathogenic submicroscopic CNVs may also occur in otherwise low-risk fetuses without ARSA.

Interpretation

- The cohort found no abnormal karyotype or pathogenic CMA findings in 82 isolated ARSA cases with low-risk screening.
- The meta-analysis showed a pooled 2% incremental CMA yield across 930 cases, but between-study variability was moderate.
- Accurate classification of ARSA as truly isolated is essential, because the presence of additional anomalies or soft markers may substantially increase the likelihood of genetic abnormalities.

Clinical message



When ARSA is truly isolated and first-trimester screening is low risk, counseling should emphasize the limited additional diagnostic yield of CMA while still supporting individualized decision-making.

Take-home conclusion

Our original cohort demonstrated no additional CMA findings in 82 fetuses with isolated ARSA and normal karyotype.

Across 10 studies including 930 cases, the pooled incremental CMA yield was low at 2% (95% CI 1–3%).

Overall, isolated ARSA in a low-risk setting appears to have a limited additional diagnostic yield for CMA, and this should guide prenatal counseling.