Background

- Rare autosomal aneuploidies (RAAs) are abnormalities in autosomal chromosomes other than 13, 18, or 21, the vast majority of which are rare autosomal trisomies (RATs).1
- Outcomes in pregnancies that screened positive for an RAT (RAT+) are not well characterized.
- Improved understanding of potential pregnancy and fetal complications resulting from RATs will help inform better prenatal care for these pregnancies.

OBJECTIVE:

- To characterize pregnancy and fetal outcomes in pregnant patients who screened positive for at least one RAT by prenatal cell-free DNA (pcfDNA) testing.

Methods

- Pregnant patients who received Prequel, a whole genome sequencing-based pcfDNA screen that incorporates fetal fraction amplification2 between 2019 and 2022 and screened positive for at least 1 RAT were included.
- Diagnosis codes, procedure codes, and pharmacy fills from Komodo’s Healthcare Map claims data were used to estimate rates of miscarriage, termination, live birth, and term versus preterm birth.

Results

- A total of 626 RAT+ pregnancies were eligible for analysis (Figure 2).
- Characteristics of pregnant patients are presented in Table 1.
- Trisomies 7, 16, and 20 were the most commonly observed RATs.

- Among 460 pregnancies with known pregnancy outcomes, 240 (52.2%) resulted in live births and 70 (15.2%) resulted in miscarriage (Figure 3).

Study limitations:

- Confirmatory diagnoses of RATs were not available in this dataset.
- Early pregnancy loss is likely underreported because this cohort only included pregnancies at 10 weeks of gestation or later.
- Capturing miscarriage and elective termination in claims data is difficult.

Conclusions

- Using a novel approach of linking pcfDNA results to insurance claims data, we observed outcomes in pregnancies that screened positive for RATs among one of the largest cohorts analyzed to date.
- RAT screen-positive pregnancies showed higher rates of preterm birth and higher rates of miscarriage as compared to previously reported general population estimates.5,6

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Poster # P-085

Pregnancy and fetal outcomes in pregnancies that screened positive for rare autosomal trisomies (RATs)

Devika Chawla1, D. Claire Miller1, Sumner Pierson1, Lyuba Popadic2, Francesca Devine3, Katie Johansen Taber3


Figure 1. Study design and data synthesis

Figure 2. Cohort selection

Table 1. Baseline characteristics of patients with RAT+ pregnancies

Table 2. Pregnancy outcomes among RAT+ pregnancies

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