

ASCERTAINMENT CATEGORY IS A PREDICTOR OF IN VITRO FERTILIZATION (IVF) OUTCOMES AMONG CARRIERS OF STRUCTURAL CHROMOSOME REARRANGEMENTS



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INTRODUCTION

- **Structural chromosome rearrangements (SR)** are a well-established **causes of infertility and recurrent pregnancy loss.**
- The impact of **ascertainment category on pregnancy and delivery outcomes for translocation carriers** has been previously documented (1,2).
- However, similar studies have not been conducted that focus on IVF outcomes for those with structural rearrangements (SR).

PURPOSE

The purpose of this study is to understand the effect of SR ascertainment on IVF outcomes.

METHOD

- **Design:** Retrospective Cohort Study
- **Setting:** Large urban academic fertility center
- **Inclusion Criteria:** All IVF cycles in which one parent has a known SR (translocation or inversion)
- **Ascertainment categories for structural rearrangement carriers:** infertility, known family history, recurrent pregnancy loss (RPL), fortuitous discovery, or unbalanced offspring.
- **Primary outcomes:** IVF outcomes including number of oocytes retrieved, mature oocyte rate (MII), fertilization rate (FERT), and blastocyst formation (BLAST) rate by ascertainment type.
- **Secondary outcomes:** characterization of usable (balanced SR without other aneuploidies) vs. unusable (unbalanced SR or had other aneuploidies).
- **Statistical analysis:** Kruskal Wallis and Mann Whitney where appropriate with significance set at $p < 0.05$.

RESULTS

FIGURE 1: ASCERTAINMENT CATEGORIES

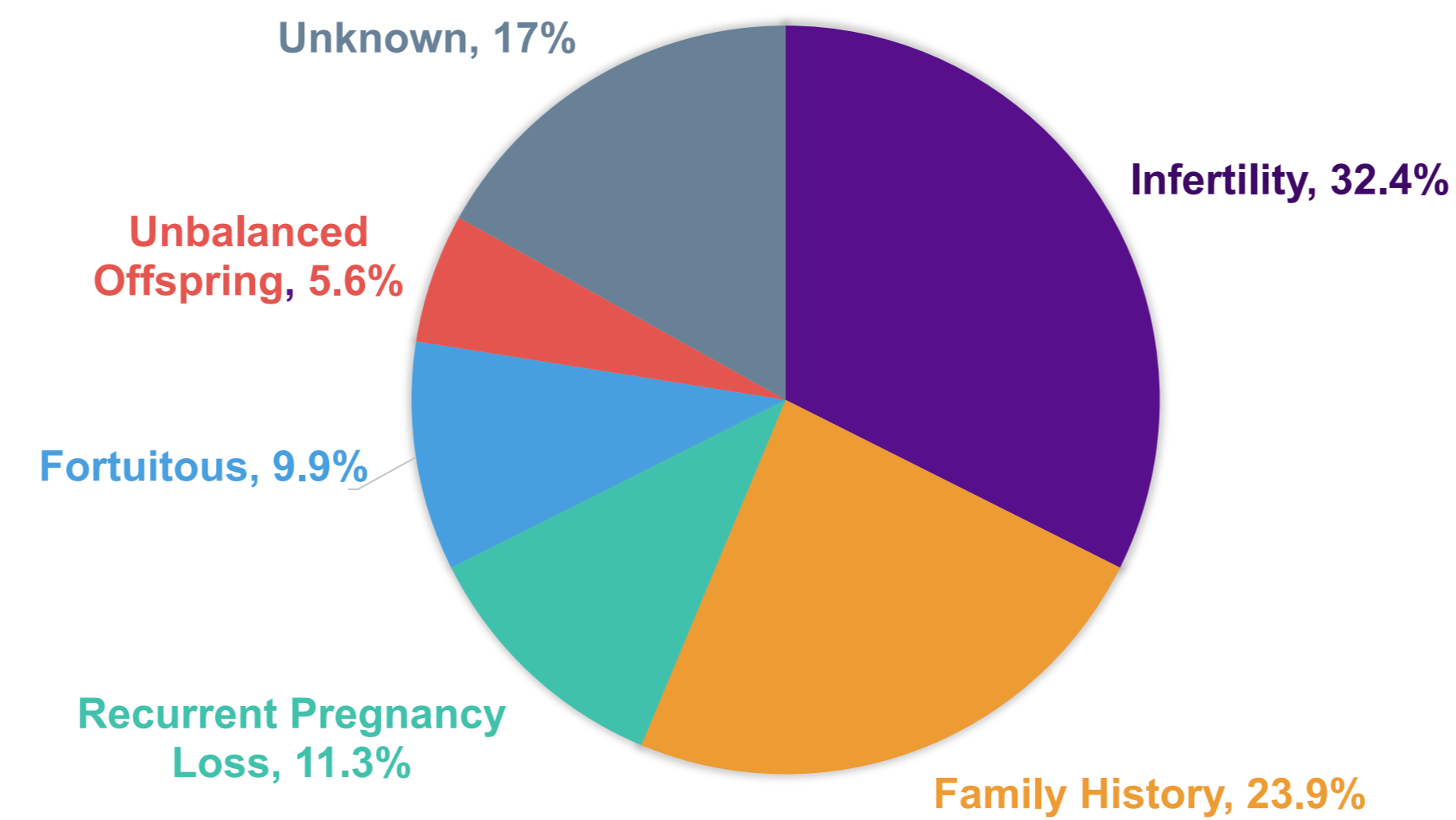


TABLE 1: IVF Outcomes by Ascertainment Category (Median [IQR])

	Infertility	Family History	Recurrent Pregnancy Loss	Fortuitously	Unbalanced Offspring	Unknown	P - Value
Oocytes Retrieved	11 [6-15.5]	18 [13.5-23.3]	11 [7.5 - 16.5]	19 [14.0 - 25.5]	20 [6.8 - 33.3]	20 [13.8 - 23.3]	<0.01
MI Rate	83.3 [73.0 - 93.6]	80.5 [64.8 - 87.7]	76.9 [69.0 - 93.6]	78.9 [68.6 - 85.0]	79.3 [63.5 - 88.5]	85.2 [77.8 - 92.4]	0.26
Fertilization Rate	73.3 [60.0 - 83.3]	71.4 [61.5 - 87.3]	70.0 [60.8 - 89.4]	70.4 [63.8 - 86.5]	75.0 [70.2 - 100.0]	80.0 [72.4 - 90.2]	0.23
Blastocyst Rate	59.2 [33.3 - 80.0]	64.3 [50.0 - 75.0]	33.3 [20.9 - 64.6]	50.0 [39.2 - 55.8]	68.6 [48.9 - 78.1]	60.6 [44.1 - 68.5]	0.03
Usable Embryo Rate	0.00 [0.00 - 21.6]	22.50 [13.5 - 33.3]	0.00 [0.00 - 21.8]	0.00 [0.00 - 30.0]	0.00 [0.00 - 18.4]	17.4 [0.00 - 37.5]	<0.01

RESULTS

- A total of 71 patients with SRs who underwent 160 IVF cycles were identified.
- Ascertainment categories are highlighted in Figure 1.
- IVF outcomes by ascertainment category are highlighted in Table 1.
- For maternal SRs alone, median oocytes retrieved remained lower among infertility (10) and RPL (10.5) categories, compared to unbalanced offspring (21), family history (18), and fortuitous (22.5) categories ($p < 0.01$).
- For paternal SRs alone, fertilization rate was significantly lower in patients with RPL (45.0%) compared to infertility (71.4%), unbalanced offspring (75.9%), family history (70.0%), and fortuitous (70.0%) categories ($p < 0.02$).
- BLAST was not associated with ascertainment category when assessing paternal translocations alone ($p = 0.051$), though this may be due to sample size given that RPL rate (median 0.0%, mean 25.0%) was seemingly lower than in other groups (median: infertility 60.0%, unbalanced offspring 72.2%, family history 66.7%).



CONCLUSIONS

- **SRs ascertained after infertility had fewer oocytes retrieved and lower rates of usable embryos** compared to SR carriers ascertained through their family history or fortuitously.
- Patients ascertained following **recurrent pregnancy loss had lower BLAST and fewer usable embryos.**
- Among paternal SR carriers whose female partners had RPL, FERT was reduced.

REFERENCES

1. Neri G, et al.. Reproductive risks for translocation carriers cytogenetic study and analysis of pregnancy outcome in 58 families. American journal of medical genetics. 1983 Dec;16(4):535-61.
2. Trunca C, et al. Reproductive risk estimation calculator for balanced translocation carriers. Current protocols. 2022 Dec;2(12):e633.

CONTACT INFORMATION

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