Time to Live Birth in Patients Who Utilize Preimplantation Genetic Testing for Structural Rearrangements

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Objective

Carriers of balanced structural chromosomal rearrangements typically show no phenotypic abnormalities yet are more likely to produce gametes with partial aneuploidies, resulting in higher incidence of miscarriage. PGT-SR identifies chromosomal rearrangements in embryos, enhancing transfer selection and pregnancy outcomes for these patient. This study aimed to assess the time from initiation of ART to FET resulting in live birth in patients who utilize PGT-SR.

Design

Retrospective cohort

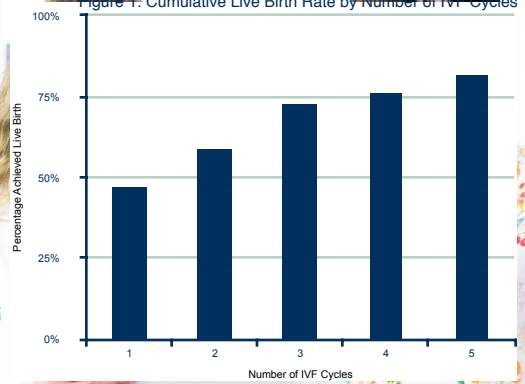
Materials & Methods

- Patients who underwent IVF with PGT-SR at a single academic center from January 2017-December 2023 were included.
- Patients were separated into groups based on live birth outcomes
- Primary outcome: time from first IVF cycle to FET resulting in live birth
- Secondary outcomes: number of IVF and FET cycles to live birth
- Student's t-test and Mann Whitney U used for analysis of continuous variables and chi-square and Fisher exact used for categorical variables. Time to live birth was modeled using Kaplan-Meier curves and adjusted survival analysis performed with Cox proportional hazards regression modeling.

Results

	All	Live Birth	No Live Birth	p
Age (mean +/- SD)	34.2 (3.7)	33.7 (3.5)	36.8 (3.8)	0.02
Type of Structural Rearrangement (N, %)				0.41
Inversion	6 (11.8)	5 (11.9)	1 (11.1)	
Microdeletion	2 (3.9)	2 (4.7)	0	
Microduplication	2 (3.9)	1 (2.4)	1 (11.1)	
Reciprocal Translocation	8 (15.7)	7 (16.7)	2 (22.2)	
Robertsonian Translocation	7 (13.7)	6 (14.3)	1 (11.1)	
Balanced Translocation NOS	25 (49.0)	21 (50.0)	4 (44.4)	
Source of Structural Rearrangement				0.72
Female Contributor	30 (58.8)	24 (57.1)	6 (66.7)	
Male Contributor	21 (41.2)	18 (42.9)	3 (33.3)	
Oocyte Maturity Rate (M2/retrieved)	77.0% (13.3)	76.5% (13.7)	79.2% (11.5)	0.58
Fertilization Rate (2pn/M2)	77.1% (14.2)	77.0% (14.6)	77.8% (13.1)	0.88
Blastulation Rate (blastocysts/2pn)	69.2% (17.6)	69.9% (17.5)	66.0% (18.5)	0.55
Biopsy Rate (biopsied/blastocysts)	67.6% (20.7)	71.1% (18.0)	51.5% (25.8)	0.0085
Euploidy Rate (euploid/biopsied)	39.2% (23.2)	41.5% (23.9)	27.2% (14.7)	0.11

Figure 1: Cumulative Live Birth Rate by Number of IVF Cycles



- Of the 51 patients who utilized PGT-SR prior to FET of a single euploid-normal embryo, 42 (82.4%) achieved [illegible]nd 9 (17.6%) did not achieve a live birth during the study per[illegible]
- Patients who achieved a live birth were younger at the start of treatment compared to those who did not achi[illegible]birth (mean age 33.7 vs 36.8, p=0.02).
- The percentage of blastocysts suitable for biopsy was significantly higher among patients who achieved a liv[illegible]1% vs 51.5%, p=0.0085).
- Among biopsied blastocysts, the euploidy rate did not differ between patients who achieved a live birth and those who did not achieve a live birth (41.5% vs 27.2%)
- IVF and FET cycle counts did not differ between groups, however patients who did not achieve a live birth spent more time undergoing treatment (median 410 vs 134 days, p=0.01)
- In a censored analysis, the median time to FET resulting in live birth was 184 days, equivalent to two IVF cycles and two frozen embryo transfers.

Conclusions

- More than half of patients who utilized PGT-SR achieved a live birth after fewer than 6 months of treatment
- Younger age and higher euploid-normal embryo counts were key predictors of success
- This study highlights the importance of PGT-SR in detecting structural rearrangements that would have been missed by other genetic testing focused solely on aneuploidy. By integrating embryologic assessment with PGT-SR, clinicians can offer more tailored guidance, empowering balanced translocation carriers with clearer pathways to achieve successful family-building outcomes.