DIFFERENCES IN IVF OUTCOMES BETWEEN PATIENTS WITH BRCA1 AND BRCA2 MUTATION

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Poster P30

BACKGROUND & OBJECTIVES

- *BRCA1* and *BRCA2* are tumor suppressor genes. Heterozygous pathogenic mutations are found in roughly 1 in 300 to 1 in 800 people, most commonly in Ashkenazi Jews.
- Heterozygous pathogenic mutations significantly increase the lifetime risk of ovarian and breast cancer, and have been associated with lower serum AMH values and earlier menopause.
- We aimed to compare IVF/ICSI, PGT-A, and PGT-M outcomes between patients with *BRCA1* vs. *BRCA2 gene* mutations.

MATERIALS AND METHODS

- Retrospective cohort study of patients with *BRCA1* or *BRCA2* mutations who underwent fertility treatment between July 2017 and October 2024.
- Cycle characteristics (age at retrieval, AMH, day 3 FSH, number of stimulation days, peak estradiol level, and total gonadotropin dose) were obtained.
- IVF/ICSI/PGT-A/PGT-M outcomes included number of oocytes retrieved, number of MII, number of 2PN, D5, D6, D7 embryos, number of biopsied blastocysts, euploid embryos, ratio of euploid embryos to biopsied blastocysts (euploid ratio), and percentage of embryos after PGT-M found to have *BRCA1* or *BRCA2* mutation (BRCA1/2 Affected).
- Incomplete/cancelled cycles and cycles with male factor (<5 million sperm/mL) were excluded.
- Performed two-tailed T-tests assuming equal variances.

Table 1: Cycle characteristics, IVF, PGT-A, and PGT-M outcomes in *BRCA1*- and *BRCA2*- positive patients

	BRCA1 (n=20)	*BRCA2* (n=19)	p-value
Age at Retrieval	33.9±4.0	32.4±4.2	0.26
AMH	2.1±1.3	3.1±2.1	0.08
Day 3 FSH	6.4±2.6	6.9±3.2	0.67
Stimulation Days	10.0±1.8	10.2±1.6	0.63
Peak Estradiol Level	1470±1666	1897±2177	0.50
Total Gonadotropins	3807±1436	3536±1531	0.58
# Oocytes Retrieved	12.4±7.7	19.3±12.9	*0.05
# MII	9.7±6.1	15.3±9.8	*0.04
# 2PN	7.3±5.4	12.2±7.2	*0.03
D5	1.4±2.1	3.8±3.8	*0.02
D6	3.0±2.7	3.7±2.9	0.47
D7	0.2±0.5	0.0±0.0	0.26
Biopsied Blastocysts	4.5±3.8	6.9±3.0	0.06
Euploid Embryos	2.3±2.2	3.2±2.3	0.25
Euploid Ratio	0.5±0.3	0.4±0.2	0.7
BRCA1/2 Affected (%)	47±36	29±24	0.11

RESULTS

- Thirty-nine cycles were included in the study; 51% (20/39) had a *BRCA1* and 49% (19/39) had a *BRCA2* mutation.
- Groups had similar cycle characteristics including FSH, number of stimulation days, peak E2 level, and total gonadotropin use. Age was slightly lower and AMH was higher in the *BRCA2* group, though this did not reach statistical significance.
- Patients with *BRCA2* had a higher number of oocytes retrieved (p=0.05), MII's (p=0.04), and 2PN's (p=0.03).
- *BRCA2* carriers reached D5 blastocyst more often (p=0.02).
- There were no differences between euploid ratio or percentage of *BRCA1*/*BRCA2* affected embryos.

CONCLUSIONS

- Patients affected by a *BRCA2* gene mutation were found to have a higher number of oocytes retrieved, MII's, and 2PN's when compared to those with a *BRCA1* gene mutation.
- *BRCA2* positive patients reached D5 blastocyst phase more often than those with *BRCA1* mutation despite similar cycle characteristics.
- *BRCA2* positive patients had a non-statistically significant lower age and higher AMH level than patients *BRCA1* gene mutations, which may have contributed to higher oocyte and blastocysts yield.

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