

## **EXPANDED CARRIER SCREEN RESULTS AND THEIR IMPACT ON IVF OUTCOMES**

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### **Background**

During in-vitro fertilization (IVF), expanded carrier screen (ECS) testing is offered to identify risk for transmitting autosomal recessive (AR) or X-linked disorders to offspring [1,2]. Though most of these tested disorders are asymptomatic in the heterozygous carrier, heterozygous status of some AR disorders can present with phenotypes, some of which can impact fertility [3].

### **Objective**

The objective was to determine if positive carrier status of disorders tested in the expanded carrier screen impacts IVF cycle characteristics and clinical outcomes.

### **Materials and Methods**

A retrospective analysis was performed using primary IVF clinic data collected by our institution. All female patients undergoing first autologous cycles from July 2019 to July 2022 with Myriad ECS, both with or without male partner ECS, were included. Outcomes of first transfer cycles through July 2023 were included.

Primary outcome was the live birth rate following the first transfer cycle using either fresh or frozen embryos. Secondary outcomes included total oocytes retrieved, fertilization rate, total number of embryos frozen or transferred, implantation rate, and miscarriage rate. Models were run using the number of positive results on the carrier screen as a continuous variable.

Subanalysis of couples with positive ECS for disorders in which carriers are known to be at risk for symptoms, as reported by Myriad, was performed.

Odds and rate ratios (OR/RR) were calculated for the number of positive ECS results and adjusted for age. Subanalysis was performed of couples with ECS positive for disorders where carriers are at risk for symptoms, as reported by Myriad. Fischer's exact and t-test were used for subanalysis.

### **Results**

A total of 217 females were found to be undergoing Myriad ECS at the time of their first IVF autologous cycle from July 2019 to July 2022. Of those, 184 had partners that also underwent ECS. We found no relationship between the number of positive results on the female ECS, or cumulative positive results of a couple, and IVF outcomes (Table). Adjustment for female age did not impact findings. Subanalysis showed no effect of carrier status on our study population.

### **Conclusions**

In this study we found that carrier status of AR or X-linked recessive disorders on ECS does not impact IVF outcomes. This is the first study looking into carrier status of disorders tested in ECS and its impact on IVF outcomes.

**Support** none

## References

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3. Rowe CA, Wright CF. Expanded universal carrier screening and its implementation within a publicly funded healthcare service. *J Community Genet*. 2020 Jan;11(1):21-38. doi: 10.1007/s12687-019-00443-6. Epub 2019 Dec 11. PMID: 31828606; PMCID: PMC6962405.

	# Positive results in female ECS					# Positive results in couple ECS						
	0	1	≥ 2	OR/RR	95% CI	0	1	2	3	≥ 4	OR/RR	95% CI
Patients (n)	98	82	37			25	62	51	34	12		
Mean Total Oocytes Retrieved	15.6	17.1	15.5	1.01	0.97 – 1.05							
Fertilization Rate (%)	70.8	71.9	70.7	0.99	0.63 – 1.56	72.1	70.8	69.2	72.5	75.6	0.98	0.67 – 1.44
Mean Embryos (frozen or transferred)	4.4	4.5	3.8	0.94	0.88 – 1.01	3.6	4.3	4.1	4.9	3.3	1.02	0.96 – 1.08
Number of Embryo Transfers (n)	91	69	31			23	55	44	30	10		
Implantation Rate (%)*	55.0	62.3	53.2	1.02	0.83 – 1.26	56.5	60.0	58.0	58.3	30.0	0.95	0.79 – 1.14

Miscarriage Rate (%)	11.3	13.0	18.8	1.16	0.66 – 2.04	14.3	8.8	11.5	16.7	0.0	1.01	0.58 – 1.78
Live Birth Rate per Retrieval (%)	45.7	48.1	36.4	0.95	0.70 – 1.29	45.8	52.7	42.0	46.9	27.3	0.89	0.68 – 1.15

\* implantation rate = fetal heartbeat/transferred embryo