



**AMERICAN SOCIETY FOR  
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**Title:**

**CLINICAL EXPERIENCE OF NURSING TEAM IN PRECONCEPTION GENETIC COUNSELING AT A LARGE, DIVERSE INFERTILITY PRACTICE**

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**Objective:**

While ethnicity based-carrier screening was once a customary component of preconception genetic testing, expanded carrier screening (ECS) is being increasingly utilized to identify recessive or X-linked mutations. The role of the nursing team has evolved to include provision of genetic counseling for patients facing new information to process and potential use in treatment. This study assesses nursing involvement and patient decision making in infertile couples accessing ECS.

**Design:**

Retrospective

**Materials and Methods:**

The study included patients who underwent ECS (panels with >200 diseases) from May 2017 – March 2018. Patients were identified as either non-carriers, carriers whose partner tested negative for the mutation, carrier couples, or female carriers (X-linked pathogenic variant). We evaluated the mutation prevalence and the decision-making process regarding use of preimplantation genetic testing for monogenic/single gene defects (PGT-M).



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## **Results:**

A total of 2439 patients (980 couples) underwent ECS. 1575 (64.6%) patients were found to carry  $\geq 1$  mutation. The most prevalent being 8.1% Alpha Thalassemia (n=198), 5.8% Biotinidase Deficiency (n=142), 5.7% GJB2-related Non-Syndromic Hearing Loss (n=139), 4.1% Cystic Fibrosis (n=100), 3.8% Familial Mediterranean Fever (n=93). 864 patients (35.4%) tested negative for all mutations.

Of 980 participating couples, 31 (3.1%) were identified as being carrier couples. 39 of the 1527 females tested (2.5%) were carriers for X-linked conditions. The 39 X-linked females and 31 carrier couples underwent formal genetic counseling to assist with ART treatment decision-making. 30 proceeded with PGT-M while 30 declined PGT-M.

Of the carrier couples who decided to access PGT-M technology, the most prevalent conditions included Cystic Fibrosis (n=5), Beta-Globin Related Hemoglobinopathies (n=4), GJB2-related Non-Syndromic Hearing Loss (n=3), Familial Mediterranean Fever (n=3), and Gaucher Disease (n=2). Of the females who pursued PGT-M for X-linked conditions, 5 were Fragile X Pre-mutation carriers and 1 a Fragile X Intermediate carrier.

Of the 30 individuals/couples who declined PGT-M, 19 were Fragile X Intermediate carriers, 7 were Fragile X Pre-mutation carriers, 2 were GJB2-related Non-Syndromic Hearing Loss carrier couples, 1 Familial Mediterranean Fever carrier couple, and 1 Beta-Globin Related Hemoglobinopathies (City of Hope variant) carrier couple. Patients positive for intermediate Fragile X were most likely to waive PGT-M.

## **Conclusion:**

Expanded genetic carrier counseling and screening have become integral parts of preconception counseling. Given decreasing costs of sequencing and increasing awareness or the discordance between self-reported ethnicity and ancestral inheritance markers, pan-ethnic is now widely utilized. The modern infertility nursing team is increasingly being called upon to provide pre- and post-test counseling to infertility patients. By educating patients about contemporary reproductive options, patients will obtain a greater sense of autonomy across their family building journey.