

Topic: Preconception Screening

ANALYSIS OF PGD UPTAKE AFTER EXPANDED CARRIER SCREENING:  
ATTITUDES AND REPRODUCTIVE DECISIONS OF HIGH-RISK COUPLES

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**Objective:** 2.20% of couples who undergo expanded carrier screening (ECS) are identified as high-risk. ECS can afford high-risk individuals additional reproductive options, including the option to pursue preimplantation genetic diagnosis (PGD). Our aim was to assess the attitudes and reproductive decisions of couples who were identified through ECS to be at high-risk of having a child with a genetic disease.

**Methods:** The study included patients identified as high-risk through ECS (couples in which both partners were carriers of the same autosomal recessive genetic disease and couples in which the female partner was a carrier of an X-linked genetic disease). Couples received a survey containing questions regarding their family planning prior to ECS, emotional reactions after ECS, reproductive decisions after ECS, and the factors which drove those decisions. Responses were analyzed and statistical analyses were performed. Informed consent was obtained.

**Results:** Based on preliminary data from 51 respondents, 44.68% of couples identified as high-risk through ECS pursued PGD. Of those who did not pursue PGD, 57.69% reported pursuing prenatal testing. Analysis showed that 93.75% of respondents reported never or rarely feeling regret about having ECS, and 77.08% of respondents reported sometimes or often feeling relieved they had ECS. 79.17% of respondents reported never or rarely having difficulty making reproductive decisions after ECS.

**Conclusions:** The near 50% uptake of PGD demonstrates the clinical impact of ECS. Of those who did not pursue PGD, one-third cited cost as the greatest prohibitive factor; as cost declines, PGD may become an increasingly accessible option. Regardless of the reproductive decision made, patients expressed satisfaction and even relief as a result of undergoing ECS. As ECS becomes an increasingly integral component of reproductive medicine, we should continue to systematically investigate its clinical utility and consider recommendations for ECS as routine care.