Genetic counselors’ perspectives on expanded carrier screening use in assisted reproductive technologies
Original research

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Introduction or statement of problem: Expanded Carrier Screening (ECS) is a pan-ethnic option to identify carriers for many heritable conditions which became clinically available in 2010. ECS is now recommended by various professional organizations, although there is limited guidance on which conditions should be included. Patients pursuing assisted reproductive technologies (ART) have been identified as ideal candidates for ECS due to the option to test embryos for genetic conditions that parents carry. The purpose of this study was to determine how frequently genetic counseling services and carrier screening availability is advertised by fertility clinics, as well as to describe genetic counselors’ experiences with the use of ECS for ART patients.

Hypothesis: Authors hypothesized that genetic counseling services are limited in the setting of ART and infertility, and that a clear pattern of counseling education strategies and panel design preferences would be identified for counseling patients in regards to ECS.

Methods/design: This study consisted of a website analysis of 462 fertility clinics which submit data to the Centers for Disease Control and Prevention National Assisted Reproductive Technology Surveillance System. This analysis recorded geographic location, affiliation status with a university or major health system, advertised availability of genetic counseling services, advertised presence of genetic counselors on staff, and availability of carrier screening. A second part of the study involved a survey of currently practicing genetic counselors, and asked participants about their experiences with patient education and panel design of ECS, as well as professional work setting.

Results: Of the 462 fertility clinics analyzed, 17% advertised carrier screening, while only 7% advertised genetic counseling services. Surveyed genetic counselors reported that they offered expanded carrier screening frequently to patients (55%, n=84) and perceived a greater ability for genetic counselors to address carrier screening counseling components when compared to non-genetic counselor providers (p<0.05 for each component). Multiple factors were perceived as important when designing ECS panels, contributing to the difficulty in determining which conditions should be included on ECS panels.

Conclusions: This research provided more context regarding the use of ECS during reproductive counseling, as well as more insight to the subspecialty of ART genetic counseling.