Introduction

Historically, carrier testing has been targeted toward specific ethnic groups, and relied on a patient’s self-reported ethnicity. In 2015, a joint statement from five societies recognized that many people do not know their exact ethnicity or may be a mix of multiple ethnicities, making it difficult to assign a single ethnicity.\(^1\) ACOG Committee Opinion 690, published in 2017, further stated “particular disorders are less likely to be confined only to a specific high-risk ethnic group because of the increasing frequency of ethnic admixture of reproductive partners.”\(^2\) Expanded genetic carrier testing allows couples to determine their reproductive risk for many autosomal recessive genetic disorders irrespective of ancestral background or geographic origin. We present a large retrospective study of carrier testing in reproductive couples to determine an estimate of at-risk pregnancies in a diverse population undergoing routine carrier testing.

Methods

- Requisitions were reviewed to identify known or potential reproductive couples.
- Progenity database was queried for names of reproductive partners, and demographic information was cross-referenced to determine matched samples.
- A retrospective analysis was performed for known matched reproductive couples and descriptive analysis regarding ethnicity, test ordered, and results of genetic carrier testing on each couple was performed.
- Risks were considered only for autosomal recessive disorders.

Conclusions

- Our study identified \(5.9\%\) of reproductive couples at increased risk to have an affected child, and demonstrates the importance of testing partners to allow for reproductive and prenatal decision-making.
- \(28\% (62\%)\) of these couples were carriers of disorders included only on expanded carrier testing panels. Broader testing increases the yield of identifying at-risk couples, and at-risk couples may be missed if carrier testing is performed based solely on reported ethnicities.
- With the continued adoption of expanded carrier testing panels, obstetrical practices should anticipate increased rates of identified carrier couples.