Evaluating the Efficacy of Three Carrier Screening Workflows Designed to Identify At-Risk Carrier Couples

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BACKGROUND

● The primary goal of carrier screening is to identify couples at-risk for having offspring with serious and prevalent genetic conditions.

● However, the strategy used for partner screening can impact the efficacy of at-risk couple (ARC) detection.

METHODS

● Three carrier screening strategies were evaluated among patients who underwent expanded carrier screening at a single laboratory (N=314,100): sequential: male partner sample collected and tested after the female partner, tandem: male partner sample collected and tested at the same time as the female partner, and tandem reflex: male partner sample collected at the same time, but only tested if the female is screen-positive (Figure 1).

● Efficacy was assessed by measuring turnaround time, partner testing compliance (testing of the male partner initially screened negative), and ARC detection.

RESULTS

● Partner compliance rates were 25.8% (sequential), 100% (tandem), and 95.9% (tandem reflex; Figure 2A).

● Overall, 42.2% of couples tested in tandem unnecessarily tested the male partner when the females screened negative (Figure 2B).

● In contrast, <4% of tandem reflex and sequential couples had unnecessary male testing (Figure 2B).

● This study demonstrates that the tandem reflex screening strategy is the most efficient way for clinics to achieve the ACOG recommendation of a standardized carrier screening approach.

CONCLUSION

● The tandem reflex screening strategy had the highest efficacy, achieving a high ARC detection rate with a short turnaround time, high partner compliance, and minimal unnecessary partner screening.

● The proportion of ARCs detected as a function of total screens was 0.5% for sequential testing and 1.3% for both tandem and tandem reflex testing (Figure 2D).