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

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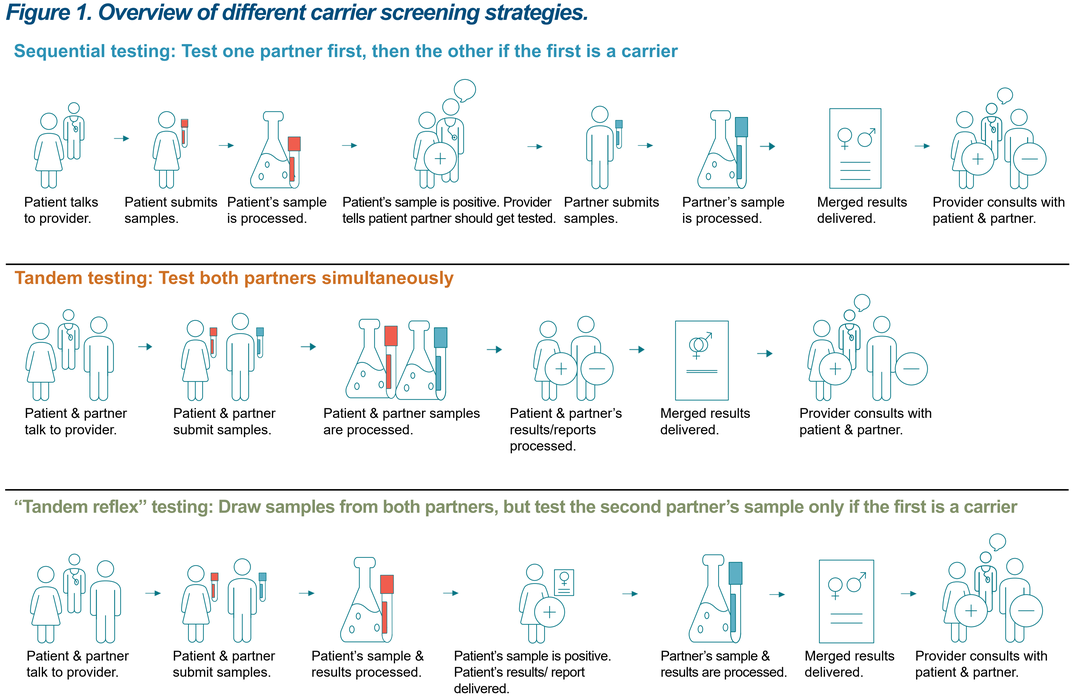
BACKGROUND

- 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 when the female partner was identified as a carrier), unnecessary testing (male receiving testing after his female partner initially screened negative), and ARC detection.

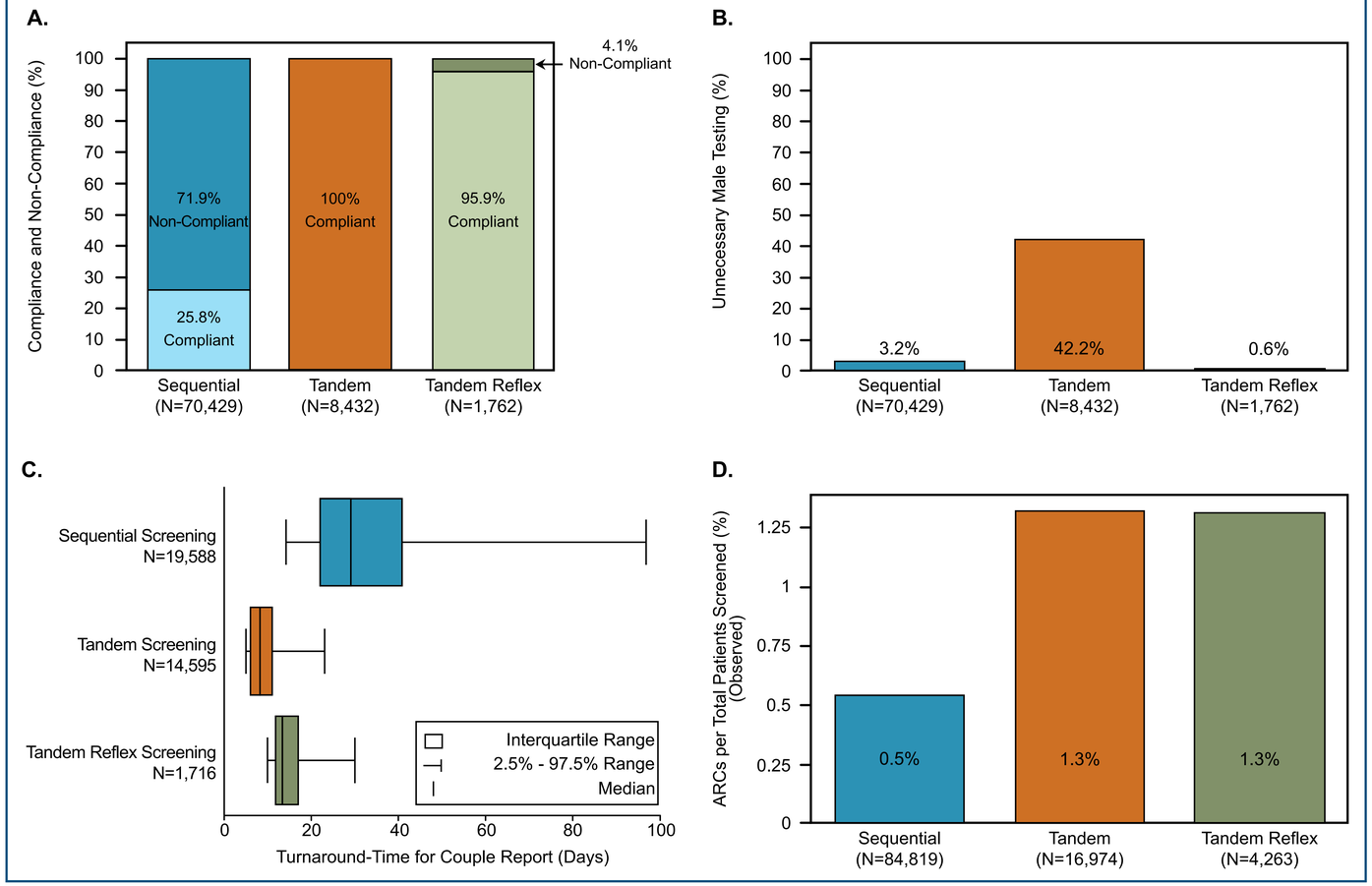
Figure 1. Overview of different carrier screening strategies.



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).
- The median turnaround times were 29.2 days (sequential), 8.0 days (tandem), and 13.3 days (tandem reflex; Figure 2C).
- 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).

Figure 2. Impact of tandem reflex strategy on ECS efficacy.



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.
- 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.