Minimizing Results Delivery Time for Couples Undergoing Carrier Screening by Using a Tandem-Submission-with-Partner-Reflex ("Tandem Reflex") Strategy

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INTRODUCTION

- Expanded carrier screening aims to detect couples at risk for having children with serious Mendelian disorders.
- As many couples are pregnant or actively trying to conceive while undergoing carrier screening, obtaining screening results in a timely manner is important.
- Reproductive partners are typically tested in a sequential manner: the female partner is tested first and, if she is a carrier, her partner is tested for the condition(s) for which she was found to be a carrier.
 - In current practice, this commonly necessitates a subsequent visit to a physician for collection and submission of the partner's sample, such that the time to receive a combined couple report is more than double the time it takes to receive an individual carrier screening report.
 - This need for a secondary sample submission imposes workflow challenges to the clinic and patients, reduces the likelihood of the partner getting screened, and may thus hamper detection of at-risk couples.
- To minimize turnaround-time and maximize the detection of at-risk couples, we implemented a "tandem reflex" strategy wherein both partners submit samples in tandem, but are tested sequentially, with the second partner's sample tested only if the first partner was found to be a carrier.
- Here, we investigated the impact of this "tandem reflex" strategy on turnaround-time and test utilization.

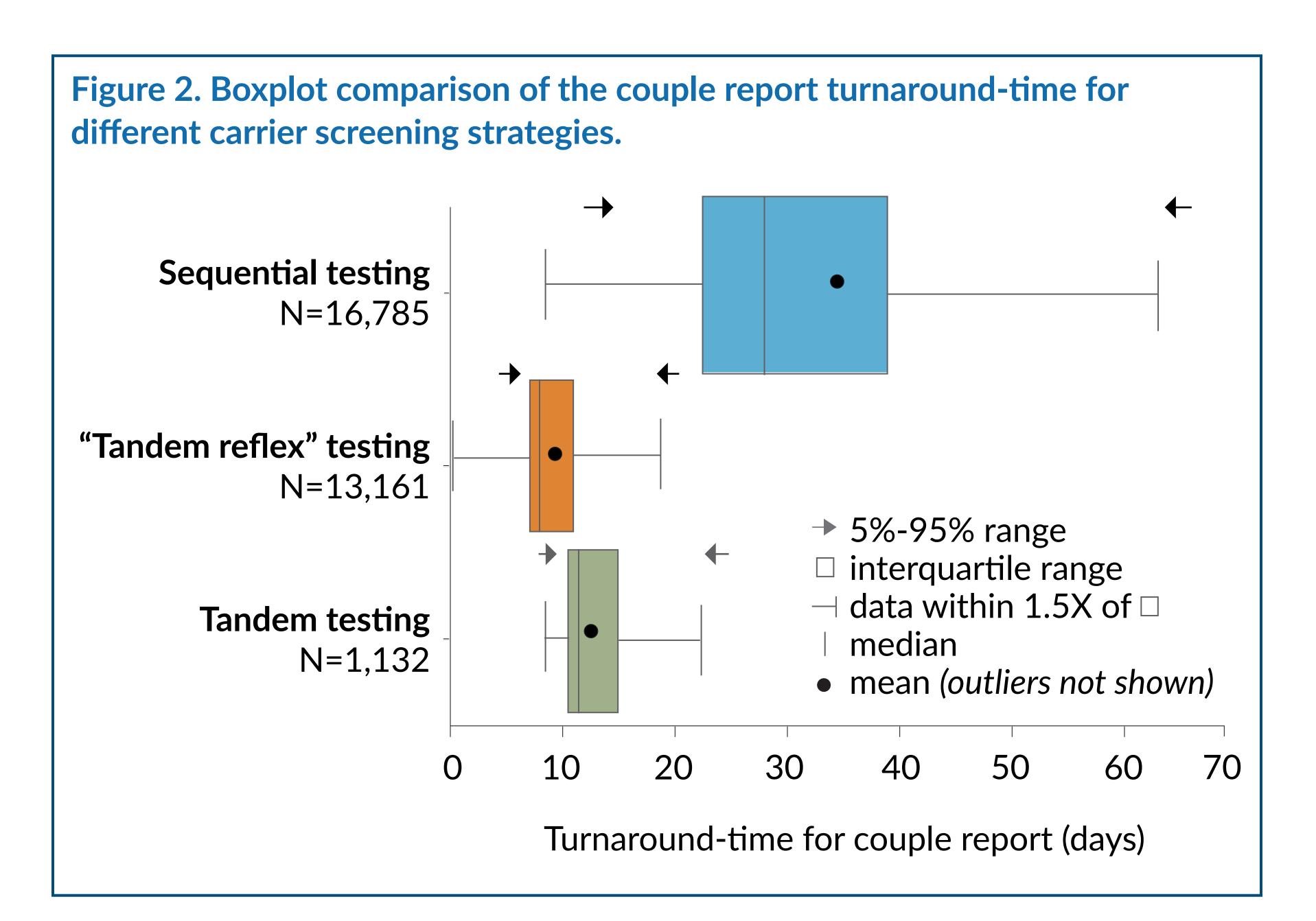
Figure 1. Overview of different carrier screening strategies. Sequential testing: Test one partner first, then the other if the first is a carrier Provider consults **Patient** Patient's Merged **Patient** Patient's sample is Partner Partner's submits submits sample is with patient & positive. Provider results sample is to provider delivered samples samples processed tells patient partner partner processed should get tested. Slower than tandem testing due to need to recontact and draw partner's sample "Tandem reflex" testing: Draw samples from both partners, but test the second partner's samp only if the first is a carrier Patient's sample Partner's sample Merged Provider Patient's Patient & Patient & partner submit is positive. consults with sample & & results are partner results Patient's results/ talk to provider patient & processed delivered report delivered partner Faster than sequential testing and cheaper than tandem testing Tandem testing: Test both partners simultaneously Patient & Patient & Patient & Provider consults Merged Patient & partner's with patient & partner submit results partner partner results/reports talk to provider delivered samples are partner processed processed Costlier than sequential testing if female is negative

METHODS

• The time between submission of a reproductive couple's first sample to the delivery of a combined report was measured before and after implementation of the "tandem reflex" strategy.

RESULTS

- Before implementing the "tandem reflex" strategy, the average time for a sequentially tested couple to receive a full couple-based carrier screening report was just under 36 days (95th percentile 75.68 days; N=16,758 couples).
- After implementing the "tandem reflex" strategy, the average wait time for a couple to receive their combined couple report was reduced to under 16 days (95th percentile just under 25 days; N=1,132 couples).
- Among 13,161 couples for which samples were submitted and tested simultaneously, 42% of the women were negative for all tested conditions.
 - In this scenario, the "tandem reflex" strategy would not have triggered testing of the male partner.
 - The average time for a couple tested in tandem to recieve results was just under 10 days (95th percentile 18 days; N=13,161 couples).
- All pairwise comparisons of the couple report turnaround-time differences are significantly different with p<0.001 (Mann-Whitney U test).



CONCLUSION

• The "tandem reflex" strategy successfully lowered the turnaround-time for receiving a combined carrier screening report by half compared to ordinary sequential testing, resulting in the receipt of information important in reproductive and pregnancy management in a more timely fashion while not adding extra costs for patients.