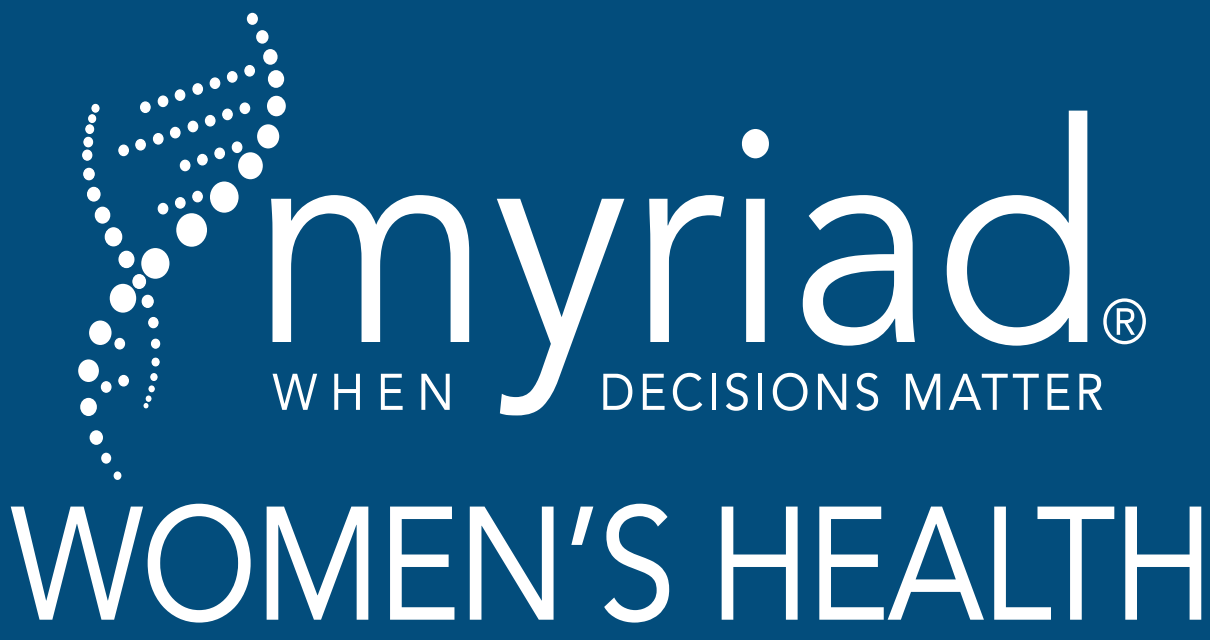


A “Tandem-Reflex” Strategy Minimizes Results Delivery Time for Couples Undergoing Carrier Screening

Aishwarya Arjunan, MS, MPH, CGC; Kristjan Eerik Kaseniit, MEng; Brandon Lee, BA; Jeff Wootton, PhD; Kenny Wong, MS, CGC; Katie Johansen Taber, PhD

Disclosure: All authors are current or former employees of Myriad Genetics, Inc. and/or Myriad Women’s Health

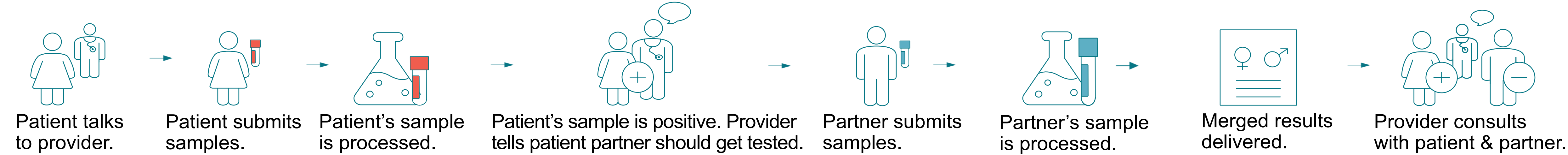


INTRODUCTION

- 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 (Figure 1).
- 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 (Figure 1).
- 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



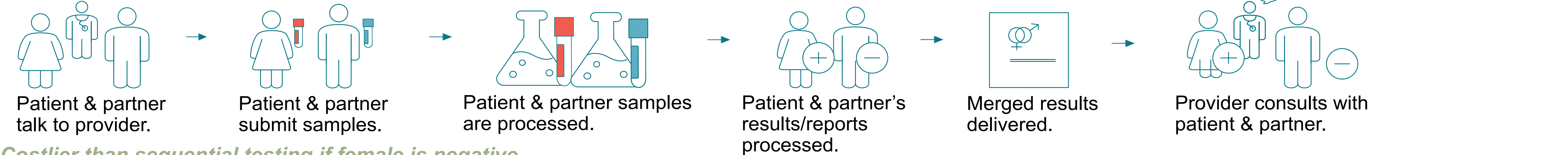
Slower than tandem testing due to need to recontact and draw partner’s sample

“Tandem-reflex” testing: Draw samples from both partners, but test the second partner’s sample only if the first is a carrier



Faster than sequential testing and cheaper than tandem testing

Tandem testing: Test both partners simultaneously



Costlier than sequential testing if female is negative

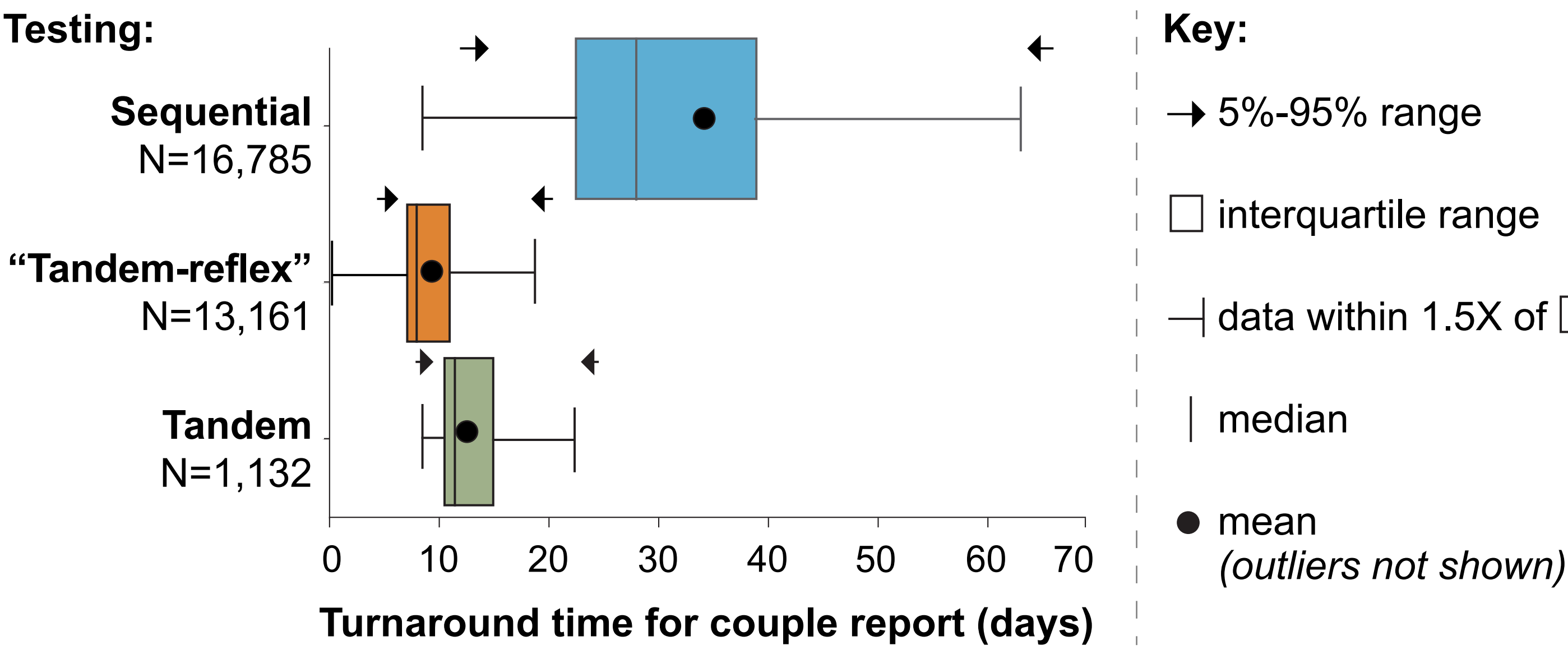
METHODS

- 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 receive 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).

Figure 2. Boxplot comparison of the couple report turnaround-time for the different carrier screening strategies



CONCLUSIONS

- 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.