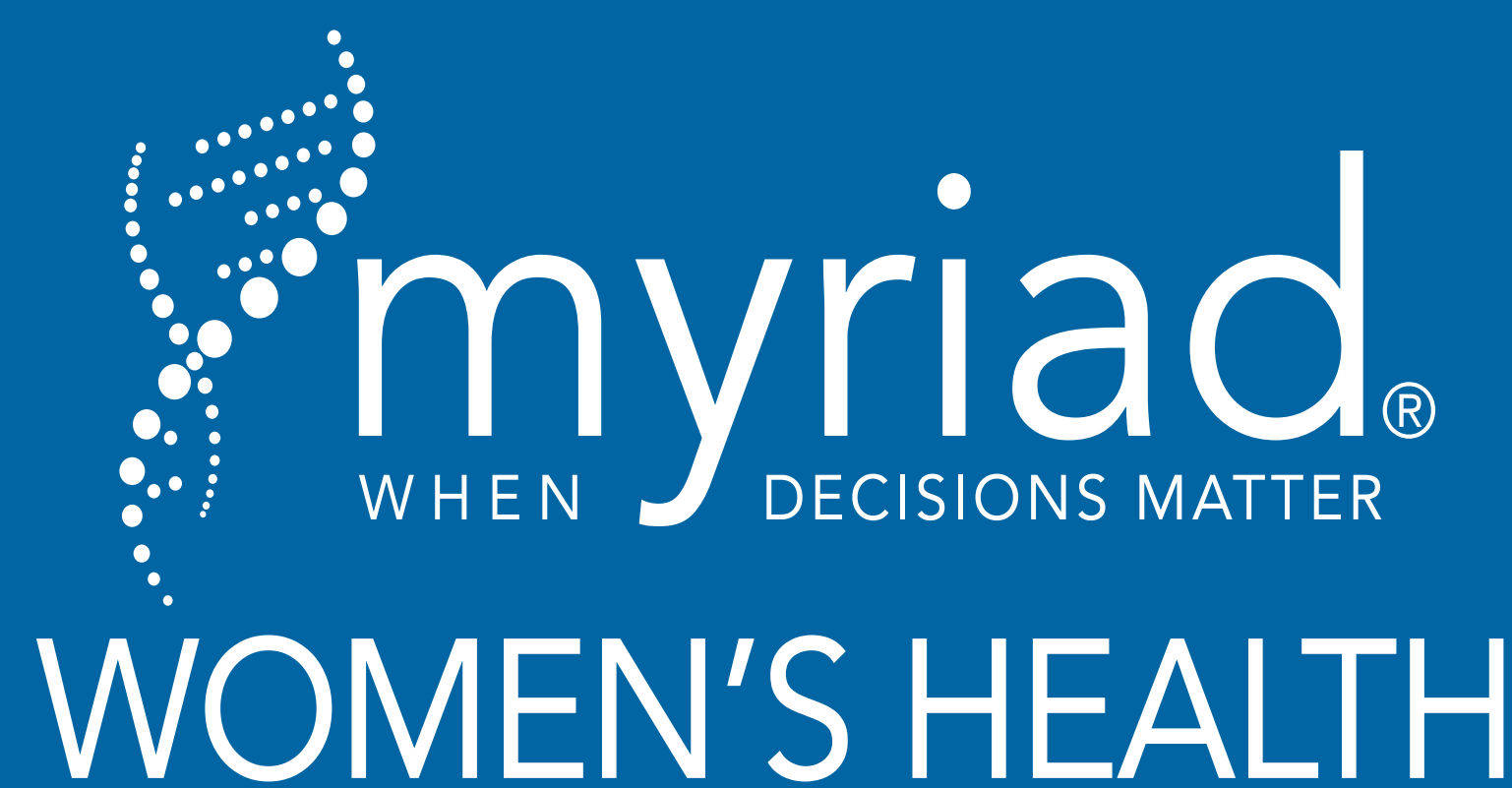


# A “Tandem-Reflex” Strategy Minimizes Results Delivery Time for Couples Undergoing Carrier Screening

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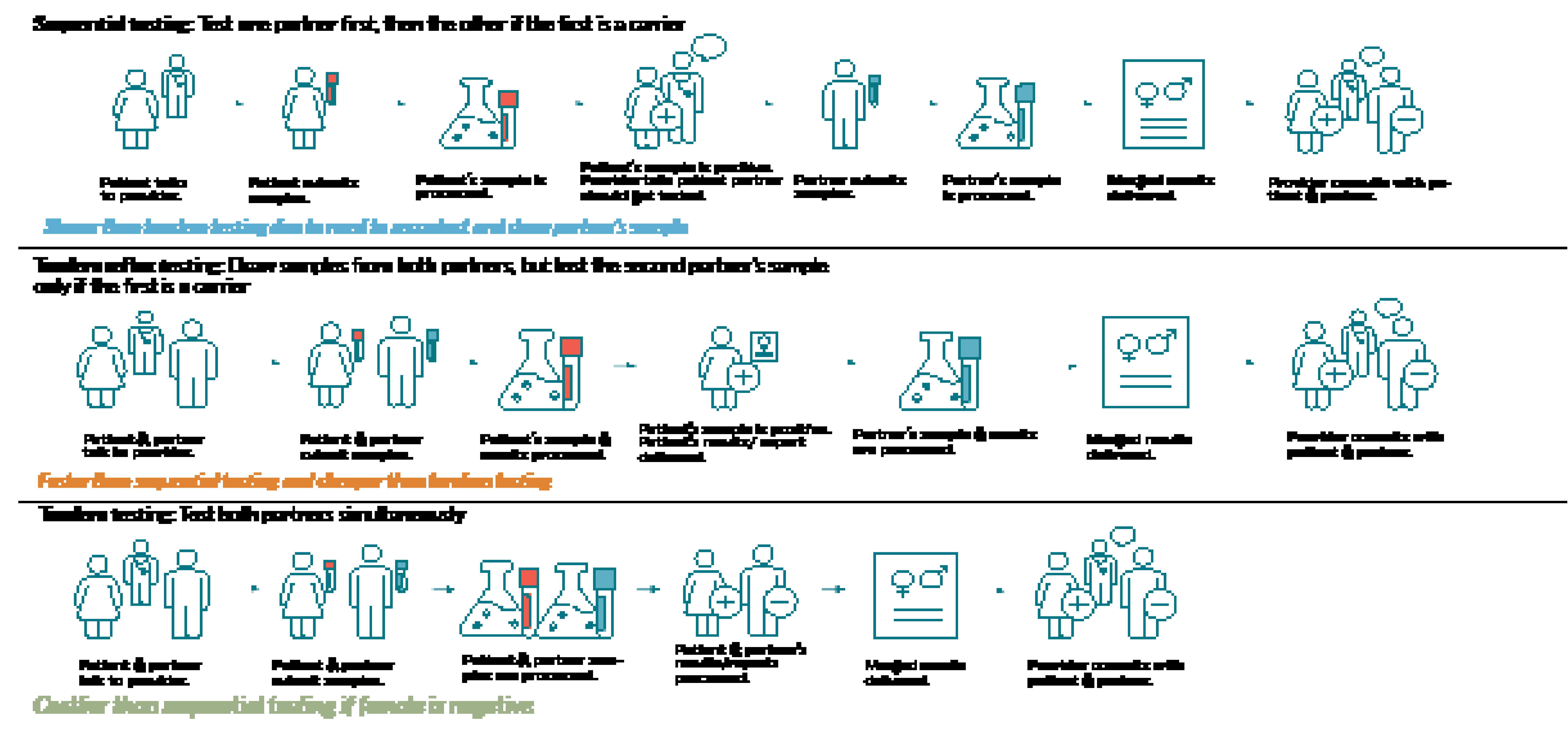
Disclosure: All authors are current or former employees of Myriad Genetics, Inc. and/or Myriad Women's Health



## 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.
  - 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



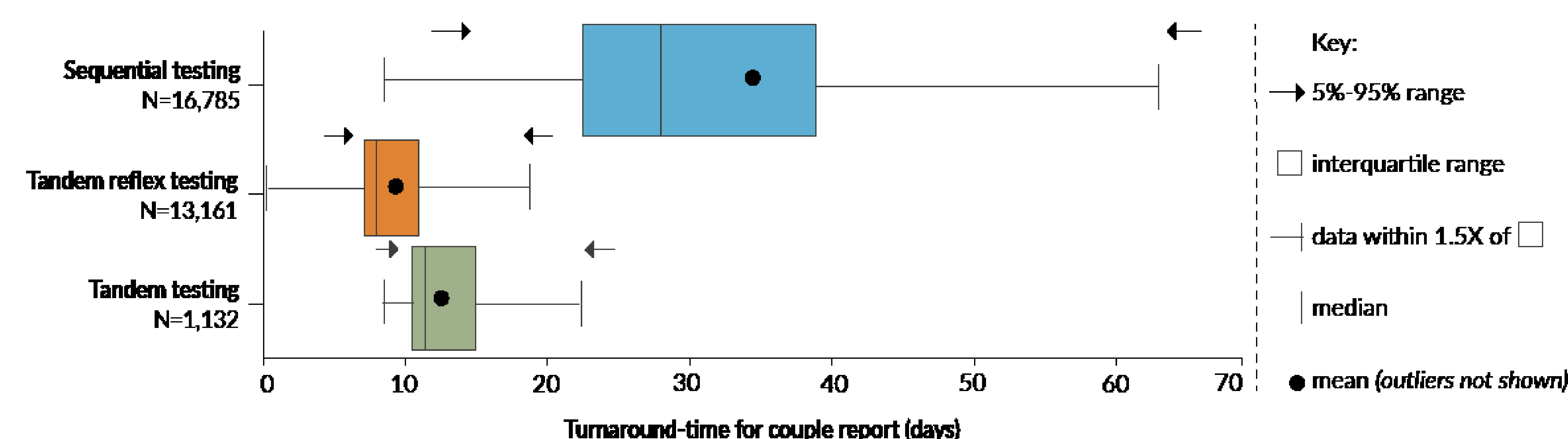
## METHODS

- 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 (95<sup>th</sup> 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 (95<sup>th</sup> 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 (95<sup>th</sup> 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 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.

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