

Geographic Disparities in Access to Genome Sequencing for Children

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Introduction

- Despite growing evidence that genome sequencing (GS) increases diagnostic yield five-fold compared with usual care (38.6% versus 7.8%), many children still undergo lengthy testing odysseys before receiving sequencing.¹
- Multiple societies have recommended GS as first-tier testing for pediatric patients with intellectual disability, developmental delays, congenital anomalies, epilepsy, and other clinical indications.^{2,3,4}
- While this guidance allows for faster diagnoses and improved care, access currently remains uneven due to limited local access to GS.

Methods

- To explore how geography and other factors might impact access to GS, we conducted a multi-state, retrospective cohort analysis on patients having recent GS at our clinical diagnostic laboratory.
- Pediatric patients from two larger states with moderate population densities, Texas (TX) and Michigan (MI), were compared to patients from two small states with high population densities, New Jersey (NJ) and Massachusetts (MA).
- Patient demographic and institutional information were reviewed.
- Distance between the patient's home zip code and their ordering institution's zip code was calculated.
- Rural-urban commuting area codes were used to establish the geographic area of each zip code.
- State rural population information was obtained from 2023 public census data.⁵

Results

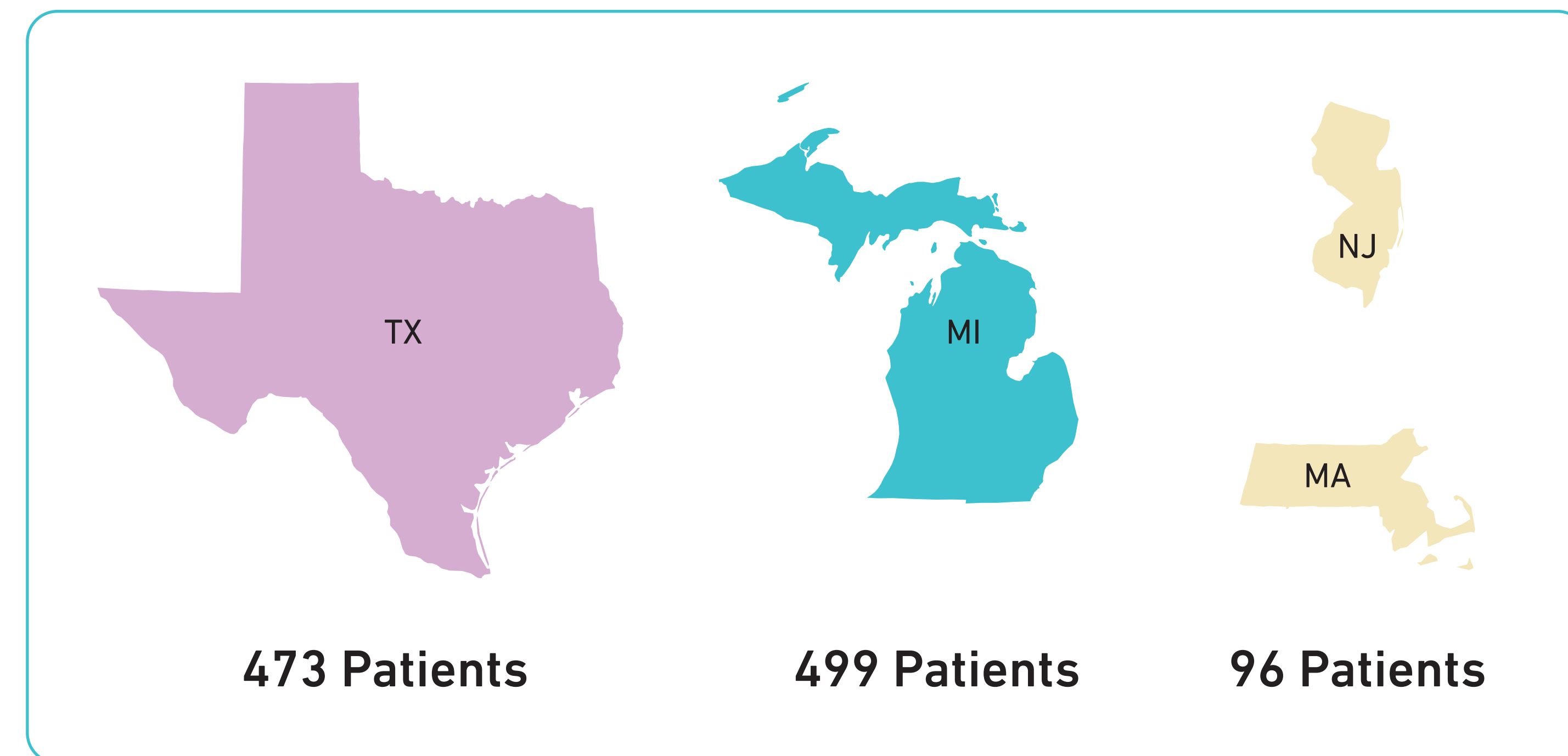


Figure 1. Distribution of patients with home addresses in TX, MI, and NJ/MA included in the retrospective cohort analysis.

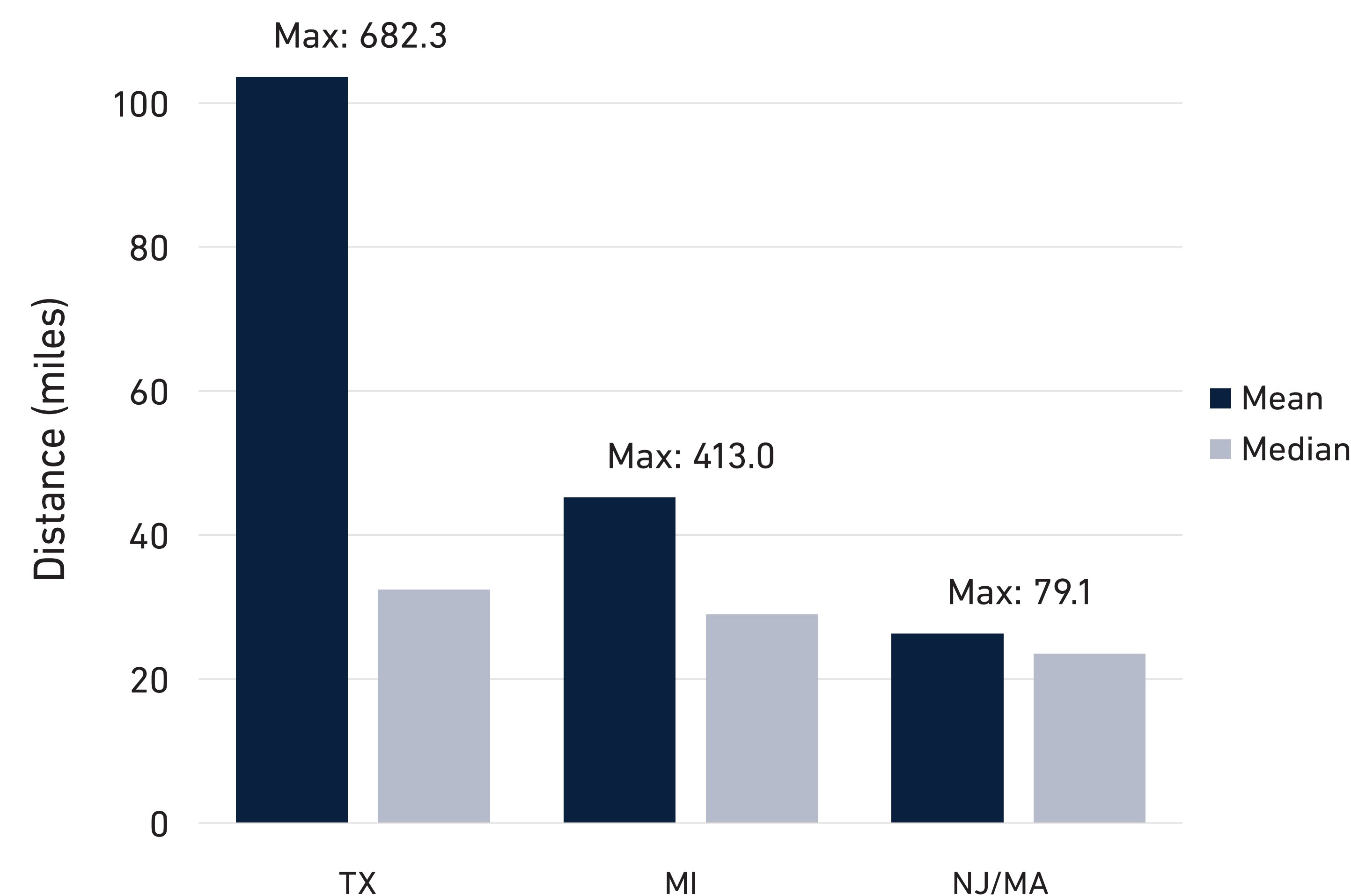


Figure 2. Comparison of mean and median distances patients lived from the ordering institution across states.

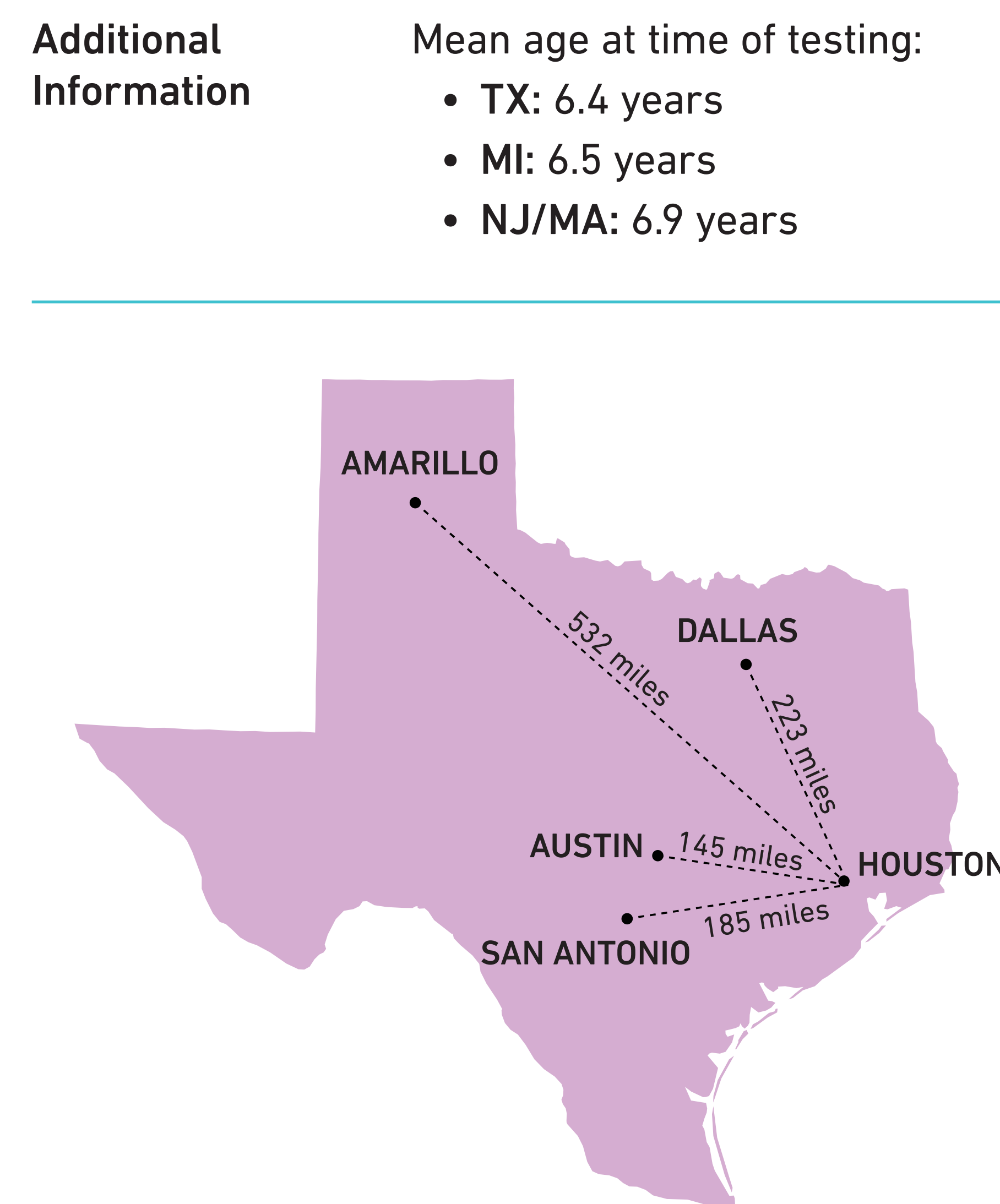


Figure 3. Approximate distances between cities in Texas to conceptualize travel distances for patients. Ninety-seven patients in TX (20.5%) and 20 in MI (4.0%) lived over 150 miles away from their ordering institution.

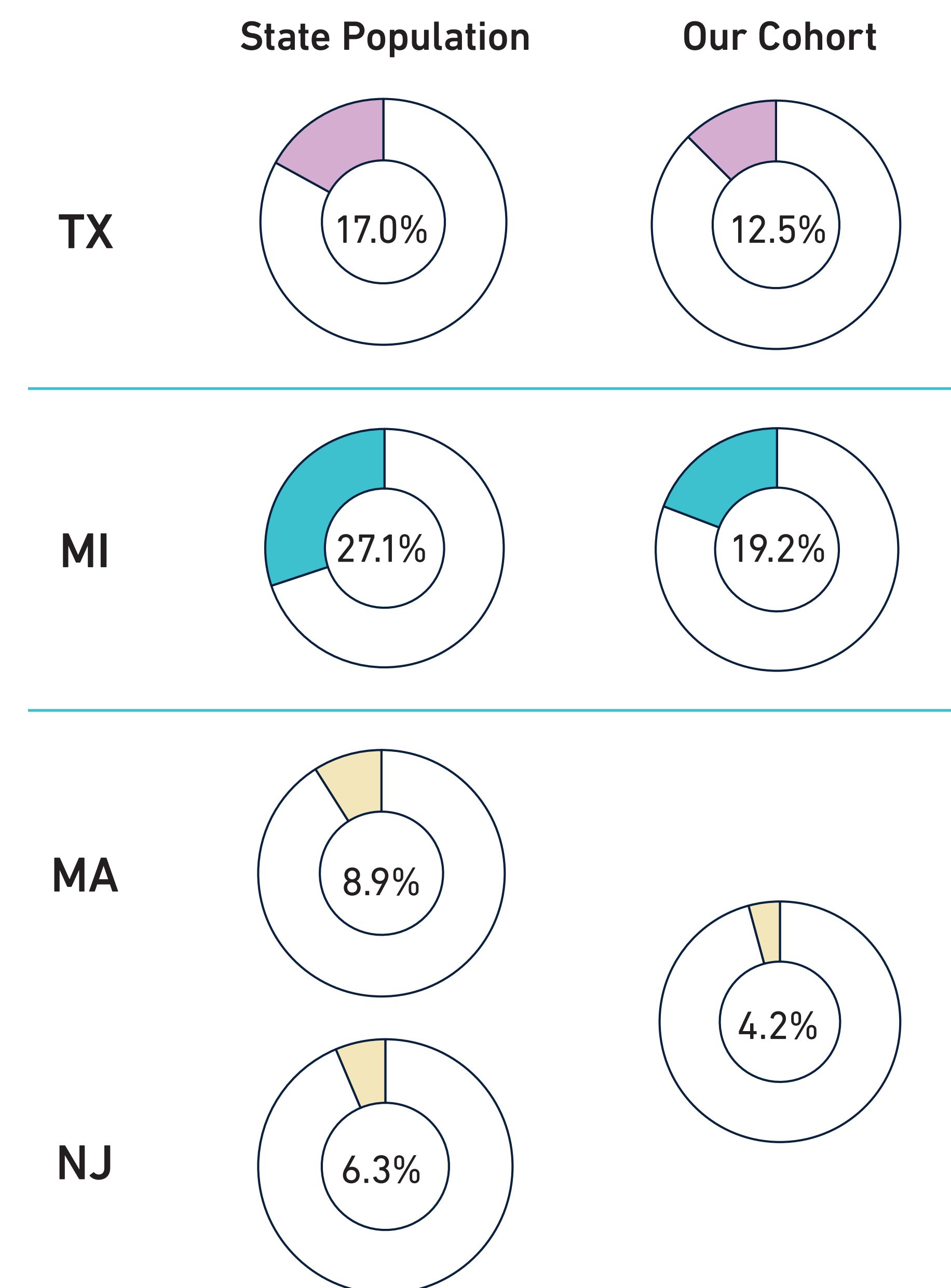


Figure 4. Percentage of patients living in rural areas compared to each state's rural population.

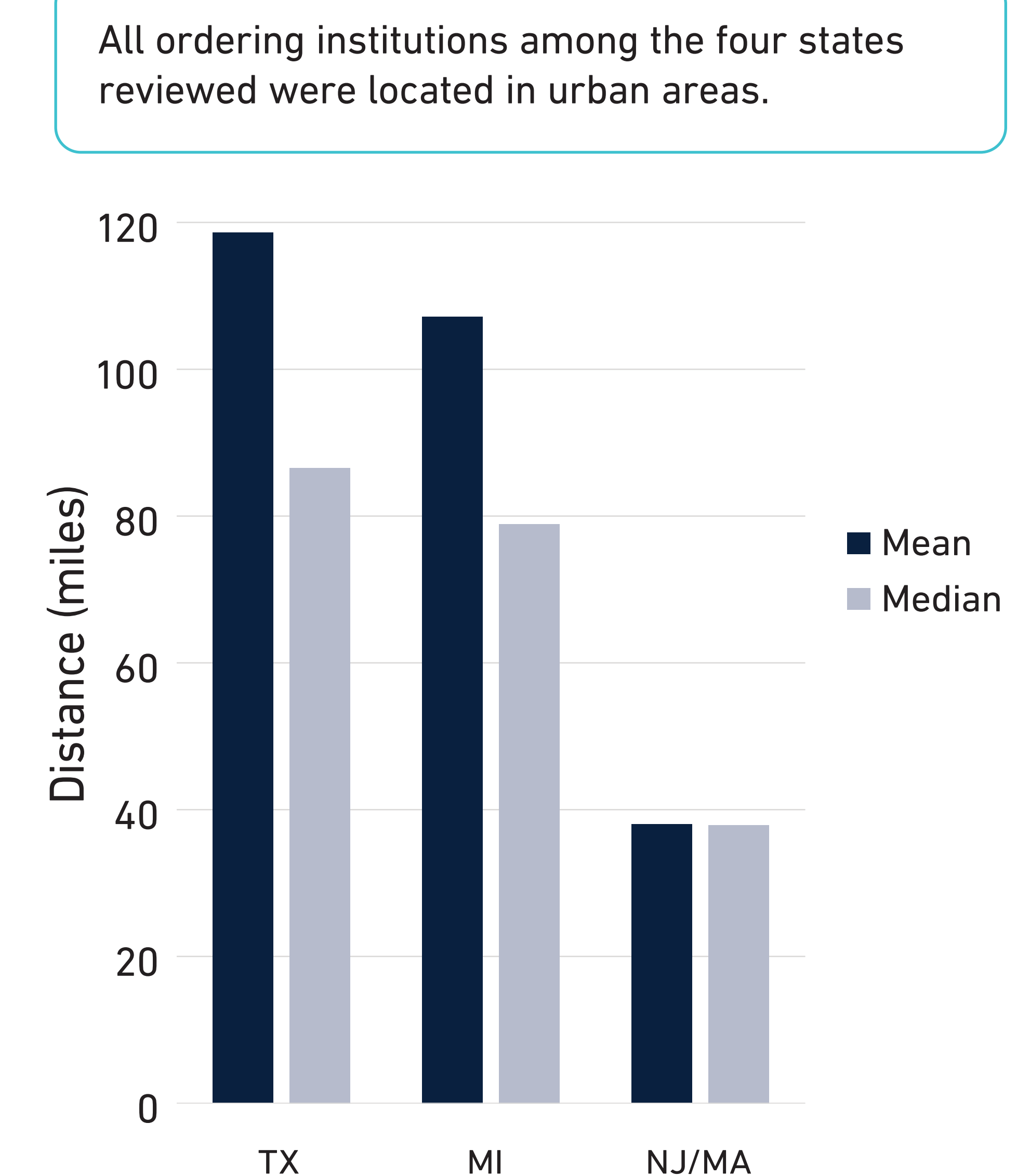


Figure 5. Difference in mean and median distances rural patients lived from their ordering institution across each state.

Conclusions

- Geographic location influences access to GS and amplifies health and financial inequities.
- Patients in TX reside farther from the institution ordering testing compared to patients in MI, as well as in more densely populated states such as NJ and MA.
- Patients living in rural areas may experience greater logistical barriers to care, especially in large states such as TX where public transit is limited outside of major cities.
- Additionally, the proportion of patients residing in rural areas is lower than the proportion of each state's rural population, suggesting patients that receive GS from rural areas may be underrepresented.
- Mobile or remote sample collection, along with increased telehealth genetic appointments, could promote access to equitable care.
- Additional studies on genetic testing access would help better understand needs.

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