Demographic and Geographic Considerations in Pediatric Genome Sequencing: A Diagnostic Laboratory Review

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Introduction

- Genome sequencing (GS) is increasingly utilized as a first-tier diagnostic tool for patients with suspected genetic disorders.
- The American Academy of Pediatrics and the American College of Medical Genetics and Genomics recommended GS as a first-tier diagnostic test for children with intellectual disability, global developmental delay, and congenital anomalies.[1,2]
- These guidelines help facilitate the early identification of genetic conditions, moving comprehensive sequencing from specialty-based testing to a standard component of pediatric evaluation.
- However, with the potential shift in ordering patterns of GS, the present utilization remains under-characterized at a population level.

Methods

- Secondary data analysis of de-identified clinical GS orders from our laboratory, encompassing pediatric patients (< 18 years old at the time of testing) regardless of clinical indication.
- The dataset included patient-level variables such as age, sex at birth, and insurance coverage.
- Distance between the patient's home address zip code and the ordering institution's zip code was calculated, and Rural-Urban Commuting Area codes were assigned to each zip code.
- Descriptive statistics were used to calculate mean and median for continuous variables.
- The Pearson chi-square (χ^2) test was applied to examine associations between categorical variables.
- The Mann-Whitney U test was used to evaluate differences in continuous variables across groups.

Results

Figure 1. Demographic and test characteristics for the cohort of pediatric patients having GS.

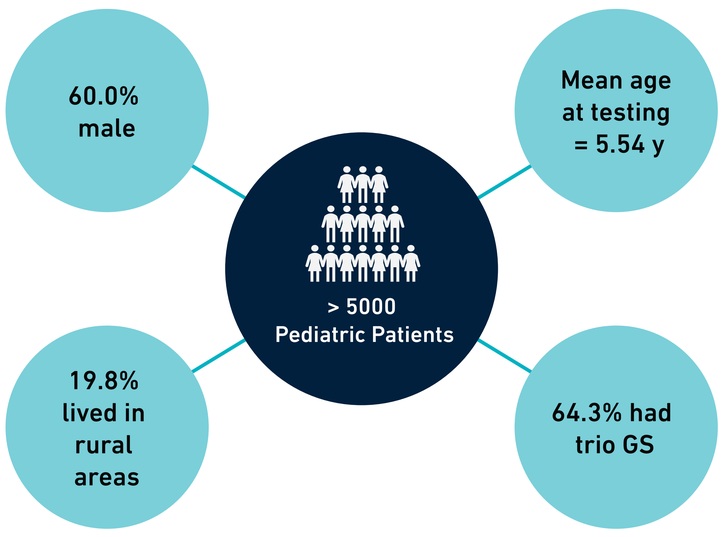


Figure 2. Contextualization of distances between patient home address and institution where genetic testing was ordered in reference to major cities around Baltimore.

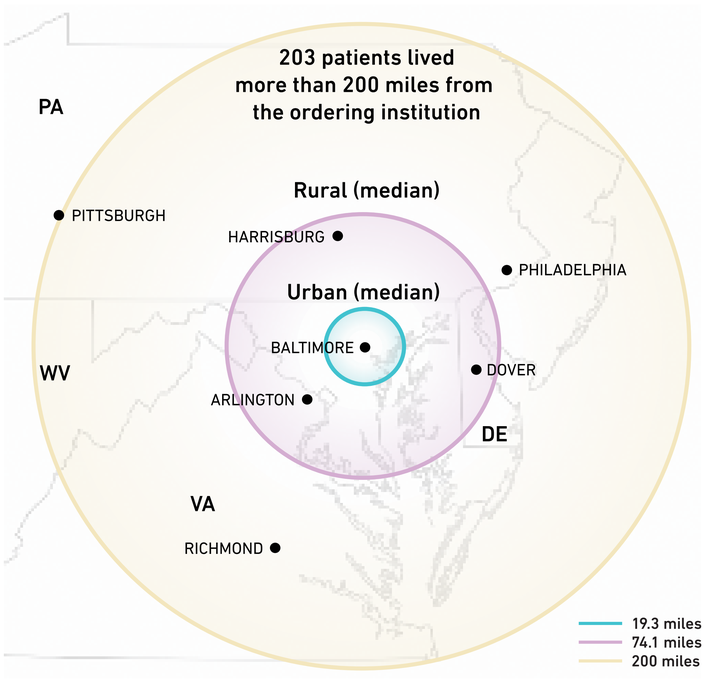
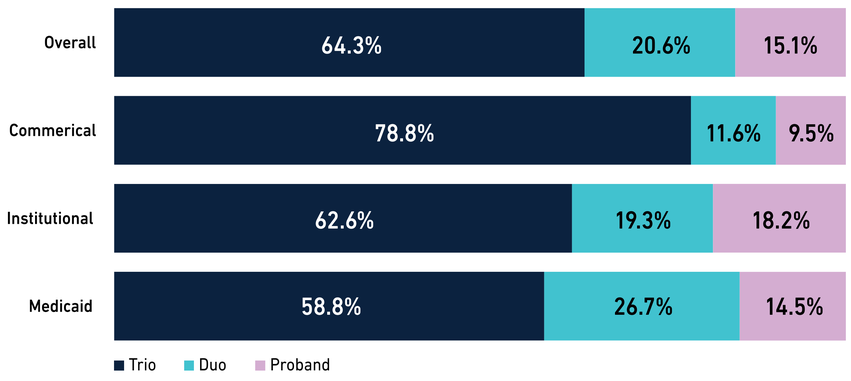


Figure 3. Test type proportions in the overall cohort and across insurance coverage groups.



Key Takeaway: Patients with commercial insurance had higher rates of trio GS than those with Medicaid or medical institution billing, with a significant difference between test type and insurance coverage (P < .001).

Table 1. Proportion of patient home addresses and ordering institutions located in rural and urban areas and patient characteristics across these geographic locations.

Characteristic	Rural	Urban
Patient home address location (%)	19.8%	80.2%
Ordering institution location (%)	2.1%	97.9%
Patient age at time of testing (mean)	5.76 years (SD= 5.3)	6.00 years (SD = 5.3)
Distance between patient home address and ordering institution (median)	74.1 miles * (M = 96.5, SD = 94.8)	19.3 miles * (M = 53.7, SD = 110.4)

* significantly different (P < .001)

Additional Facts

- The mean distance a patient lived from the institution where genetic testing was ordered was 62.0 miles (SD = 108.9), with a median of 28.6 miles.
- There were 440 patients who lived in a different state than the ordering institution.
- Almost all rapid GS were through hospital billing (98.0%).
- Payer source did not differ significantly between rural and urban groups.

Conclusions

- GS is guideline-recommended as a first-tier test for a number of clinical indications and has been shown in previous studies to help resolve the diagnostic odyssey.
- Expanding access to comprehensive GS — including trio and rapid testing — within Medicaid and commercial programs across the U.S. would likely increase patient access to timely genetic diagnoses.
- We observed significantly longer travel distances for testing for rural patients which impacts equitable access.
- Further, longer travel distances create added financial and practical barriers, including lost wages, lodging costs, and meal expenses, which can be prohibitive for many families, especially those using Medicaid.
- Equal access to GS can be supported with additional testing pathways, such as mobile phlebotomy and telehealth genetic appointments, are made available to patients living in rural communities.

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References:

1. Manickam K, McClain MR, Demmer LA, Biswas S, Kearney HM, Malinowski J, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021 Nov;23(11):2029–37.
2. Rodan LH, Stoler J, Chen E, Geleske T, Council on Genetics. Genetic Evaluation of the Child With Intellectual Disability or Global Developmental Delay: Clinical Report. Pediatrics. 2025 Jun 23;e2025072219.