Post-Implementation Evolution of Genetic Testing Pathways Following Texas Medicaid Coverage of Genome Sequencing with a Focus in Pediatric Neurology

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Background

- Limited insurance coverage of genome sequencing (GS) has been a barrier to equitable access for children with suspected rare diseases, leading to prolonged diagnostic odysseys and involvement of sequential testing strategies.
- GS has demonstrated higher diagnostic yields than exome sequencing (ES) and other testing approaches[1,2] and has been recommended as a first-tier test for certain clinical indications since 2021.[3]
- On September 1, 2024, Texas Medicaid implemented coverage for GS for children with suspected rare diseases, including neurologic indications.
- This study characterizes temporal changes in GS testing patterns among pediatric neurology patients following implementation of the policy.

Methods

- We conducted a retrospective analysis of Texas Medicaid beneficiaries less than 18 years of age who received GS after policy implementation.
- Cases were included if the child had a clinical indication of intellectual disability or developmental delay (ID/DD).
- Three consecutive 6-month post-implementation periods (early, mid, late) were evaluated.
- Ordering provider specialty and genetic testing history were abstracted from clinical documentation.
- Chi-square tests were used to evaluate differences in testing trends across the three time periods.

Results

Figure 1. Proportion of GS tests ordered by non-genetics providers or clinics across the three post-implementation periods.

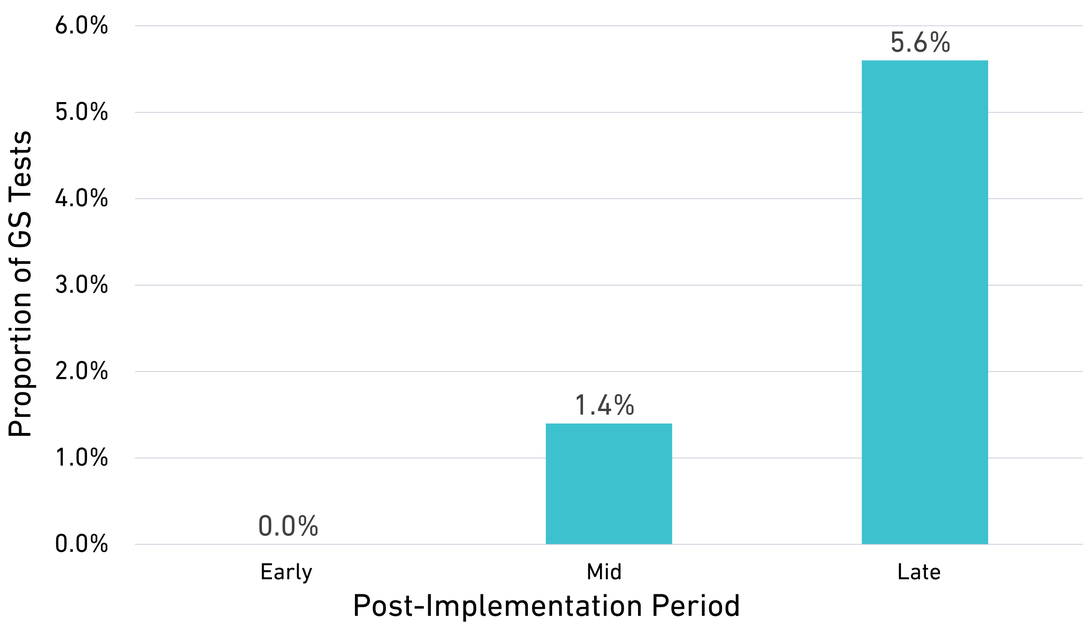


Figure 2. Proportion of children who had genetic testing prior to undergoing GS across the three post-implementation periods.

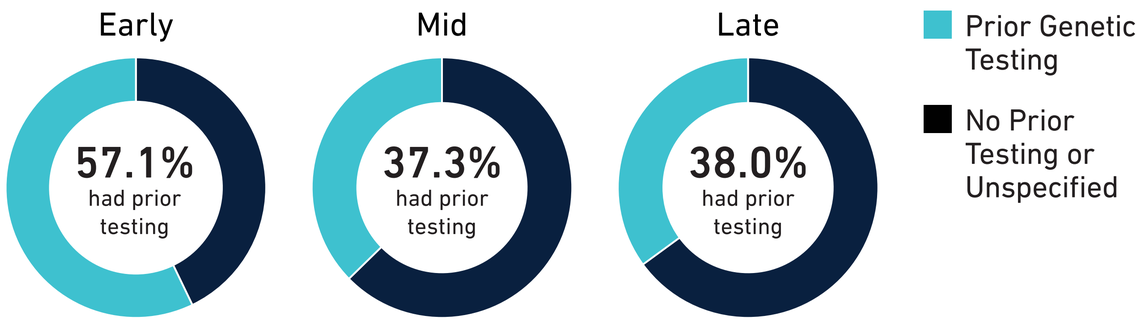


Figure 3. Proportion of children with prior genetic testing who underwent ES before GS across the three post-implementation periods. A significant difference was observed ($\chi^2 = 6.7$, $p < .05$).

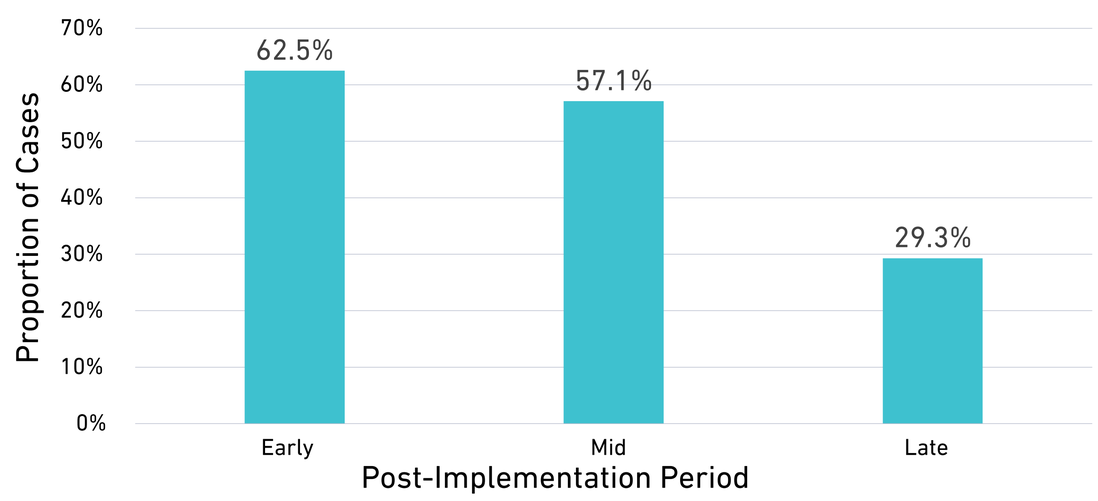


Table 1. Proportion of children who had genetic testing prior to GS that had each type of genetic test.

Test	Proportion of Cases
Chromosomal Microarray	53.2%
Exome Sequencing	42.9%
Multi -Gene Panel	41.6%
Fragile X Repeat Expansion	15.6%
Single Gene Testing	11.7%
Karyotype	10.4%
Mitochondrial Genome Assessment	6.5%

Key Findings:

- The proportion of GS tests ordered by non-genetics providers **increased** across the three periods.
- The proportion of children who had any genetic testing prior to GS **decreased** after the early post-implementation period and remained stable during the mid and late post-implementation periods.
- Further, the proportion of children with prior genetic testing who underwent ES before GS **decreased** over time.

Conclusion

- Results suggest ongoing post-policy maturation of clinical practice, with increasing utilization of GS for children with ID/DD in non-genetics clinics and as an earlier diagnostic test rather than a stepwise escalation after other types of genetic tests.
- The observed decline in cases with ES prior to GS also may suggest growing confidence in GS as a first-line comprehensive genomic testing strategy.
- The observed trends would be expected to continue over time, resulting in shorter diagnostic odysseys, fewer genetic tests, and accelerated clinical management changes with the use of GS as a first-tier test for children with ID/DD.

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References:

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3. Manickam K, McClain MR, Demmer LA, 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;23(11):2029-2037.