

Patterns in Genome Sequencing Utilization Among Texas Medicaid Children with Intellectual Disability and Developmental Delay

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Introduction

- In 2021, the American College of Medical Genetics and Genomics published clinical guidelines which recommended that genome sequencing (GS) and exome sequencing (ES) should be considered as first-tier tests for children with intellectual disability and developmental delay (ID/DD).¹
- Compared to ES or other traditional testing approaches, GS enables a more comprehensive analysis of multiple variant types within a single test, allowing for higher diagnostic yields.^{2,3}
- More recently, health insurance policies, including Texas Medicaid, have started to include coverage of GS for specific clinical indications such as ID/DD.
- The objective of this research is to examine patterns in the utilization of GS for children with ID/DD, including age at testing, prior genetic testing history, and diagnostic yield, following the implementation of GS coverage through Texas Medicaid policy.

Results

Figure 1. Number of children with ID/DD who had Texas Medicaid and underwent GS across three post-policy implementation periods.

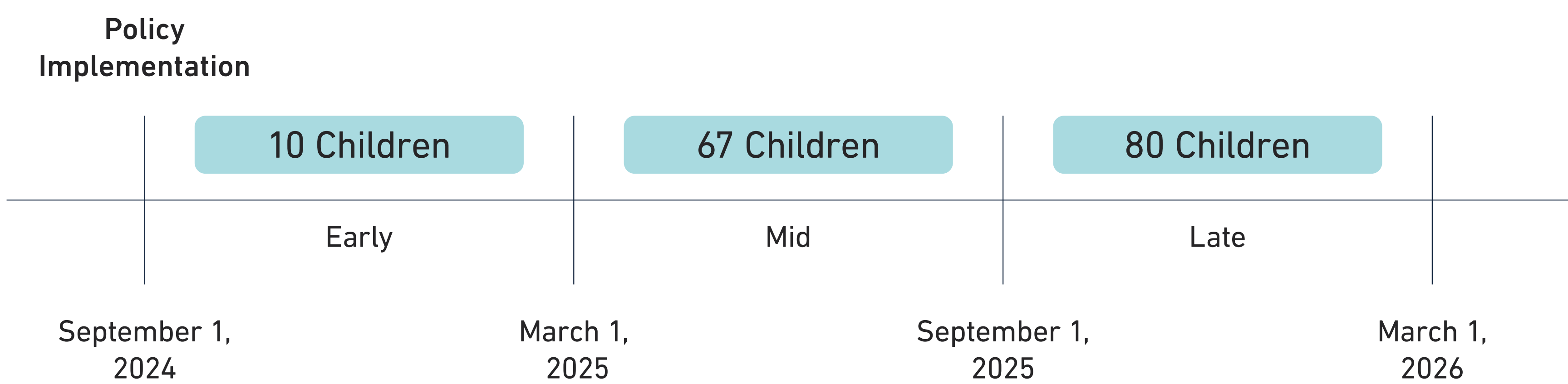


Figure 2. Proportion of children who had genetic testing prior to GS and those who had GS as a first-tier genetic test.

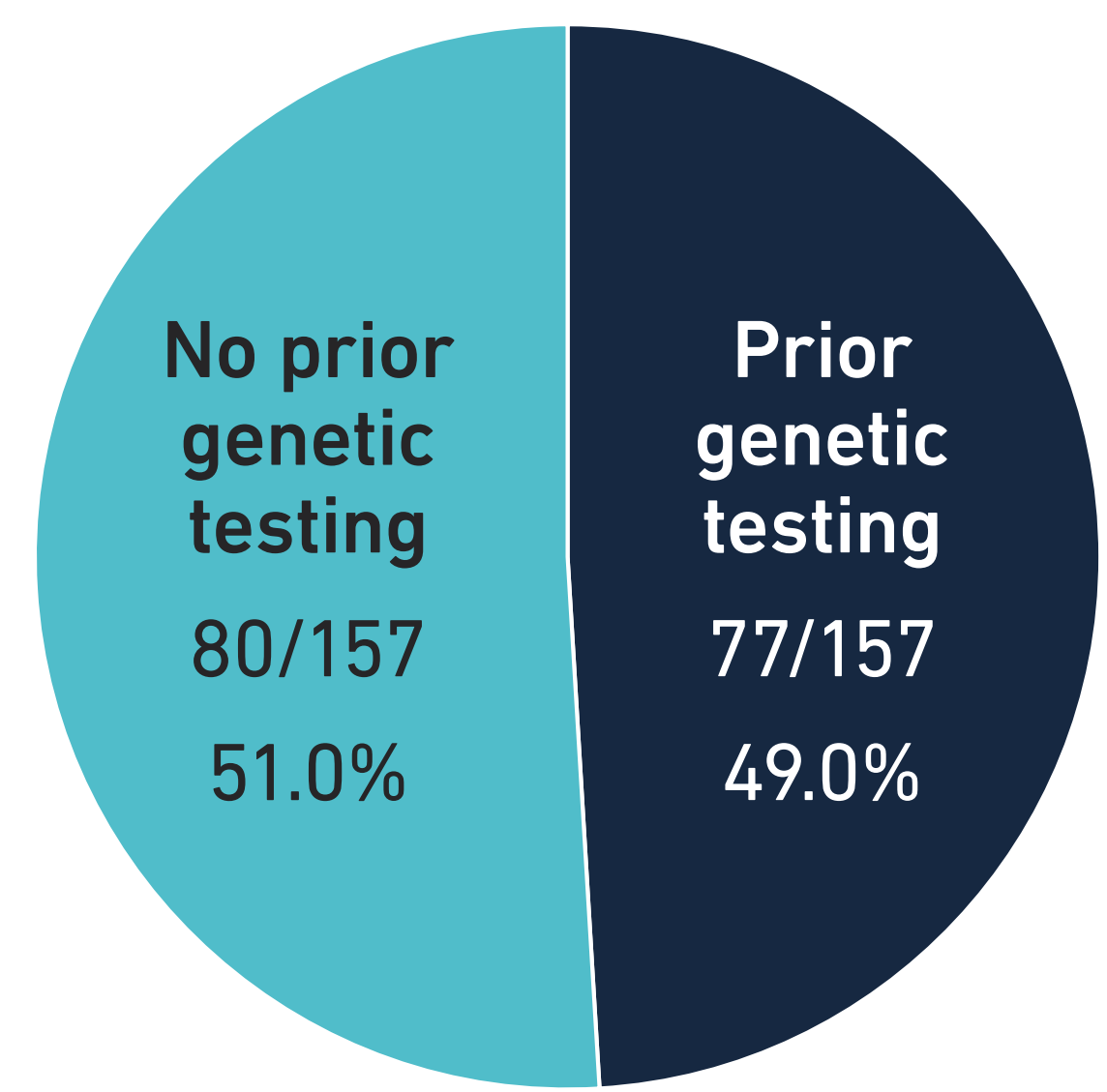
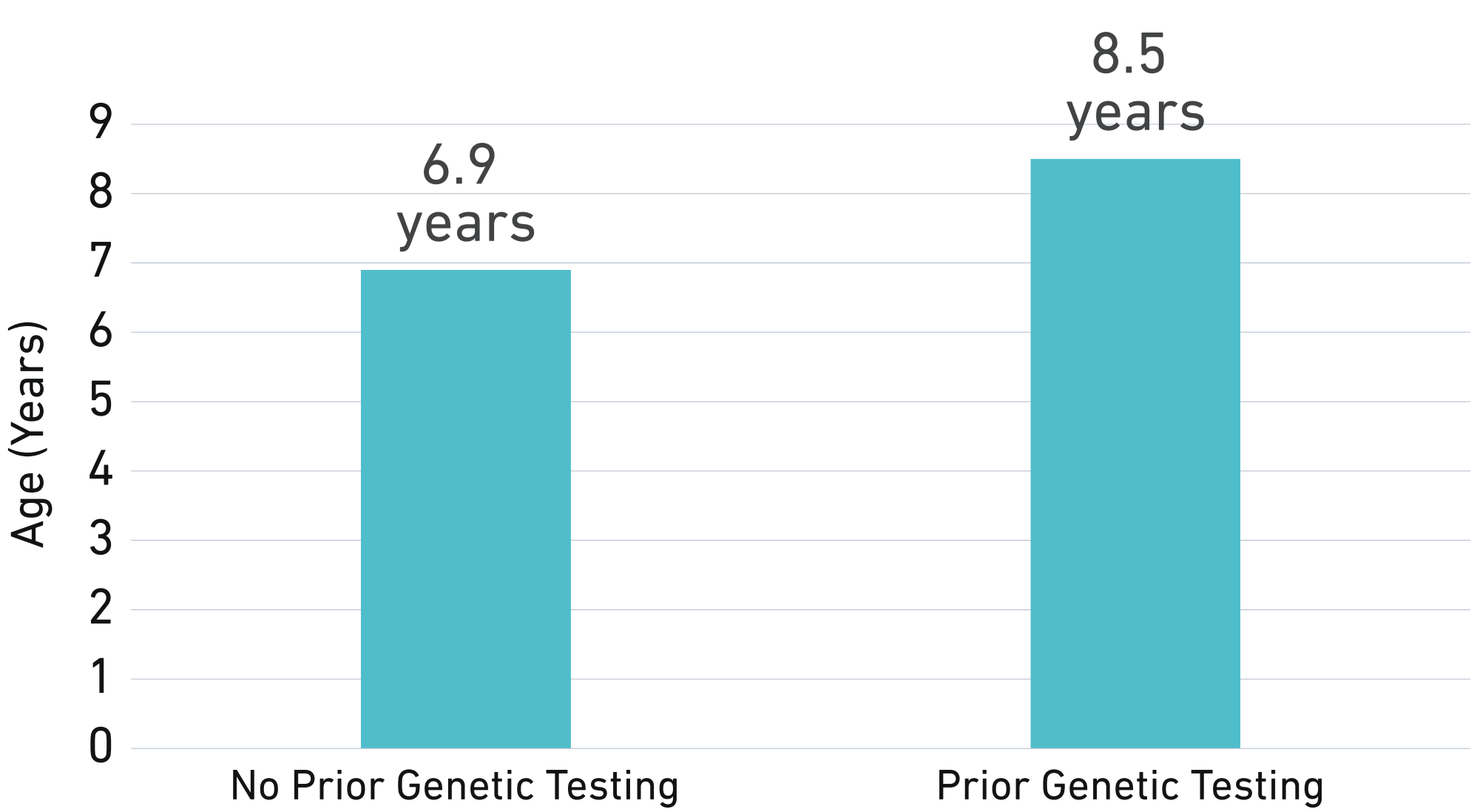


Figure 3. Mean age at time of GS for children with and without prior genetic testing.



Children with no prior genetic testing were significantly younger at time of GS compared to children with prior testing ($p < .05$).

Table 1. Number of children who were documented to have each genetic test prior to GS.

Test	Number of Cases
Exome Sequencing	33
Chromosomal Microarray	41
Multi-Gene Panel	32
Fragile X Repeat Expansion	12
Mitochondrial Genome Assessment	5
Other (karyotype, single gene, etc.)	23

Note: Some cases involved multiple panels or multiple tests within the Other test category.

Figure 4. The proportion of children with prior genetic testing who underwent ES before GS significantly decreased across the three time periods ($p < .05$).

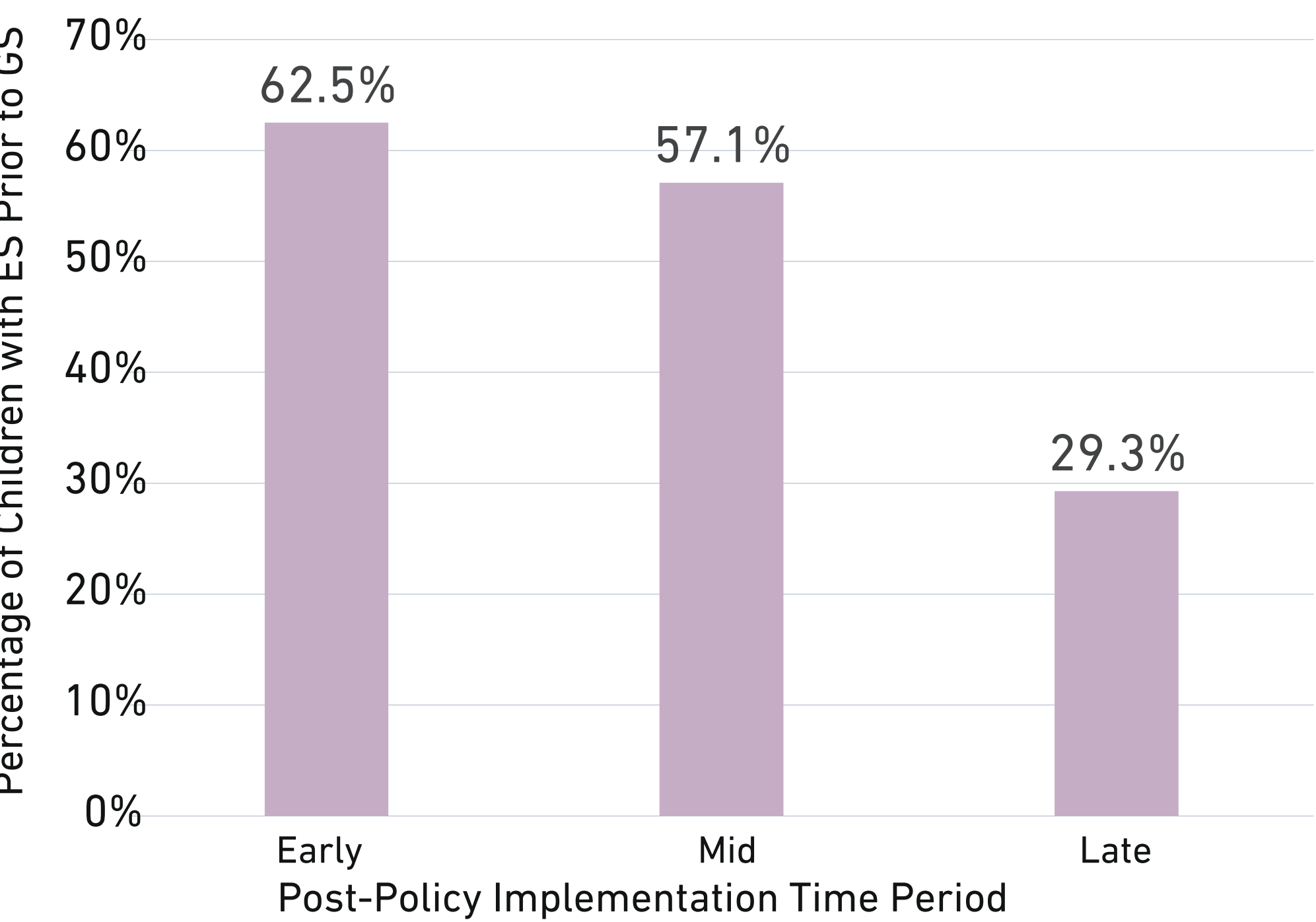
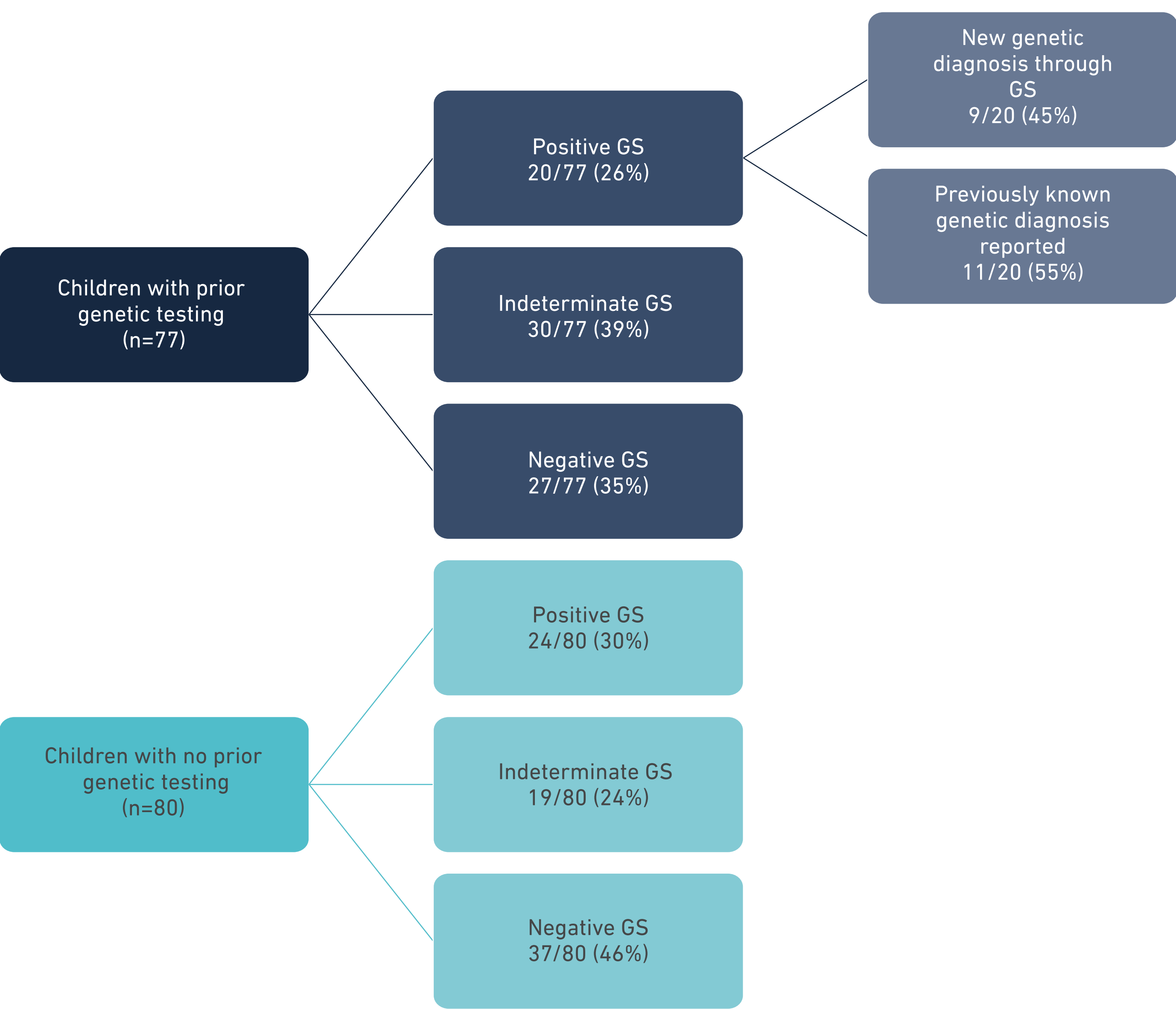


Figure 5. Results of GS for children with ID/DD across those with and without prior genetic testing.



Among those with prior genetic testing, 11.7% had a new genetic diagnosis by GS that had not been established by previous genetic testing.

Conclusions

- Among Texas Medicaid-insured children with ID/DD, the likelihood of having no prior genetic testing before GS appears to be influenced by age, suggesting differential diagnostic opportunity rather than access gaps alone.
- The decreasing proportion of children in this cohort having ES before GS aligns with the Medicaid policy, as well as the enhanced identification of variants through GS compared to ES.
- These findings support earlier, proactive genetic testing strategies by providers with Texas Medicaid-insured children to mitigate age-related diagnostic delays, reduce prolonged diagnostic odysseys, and improve care coordination for children with ID/DD.

References:

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