

Background

- Nationwide implementation of genomic newborn screening (gNBS) will require timely and high-volume return of results.
- Existing care delivery models may not be able to meet anticipated volume.
- Nationwide high-throughput telehealth genetic counseling is one care model that may be able to meet return of results and initial counseling needs.

Aim

Investigate the potential for a scaled nationwide telehealth genetic counseling service to provide timely return of results for gNBS

Methods



Retrospective chart review



November 2019 to February 2025

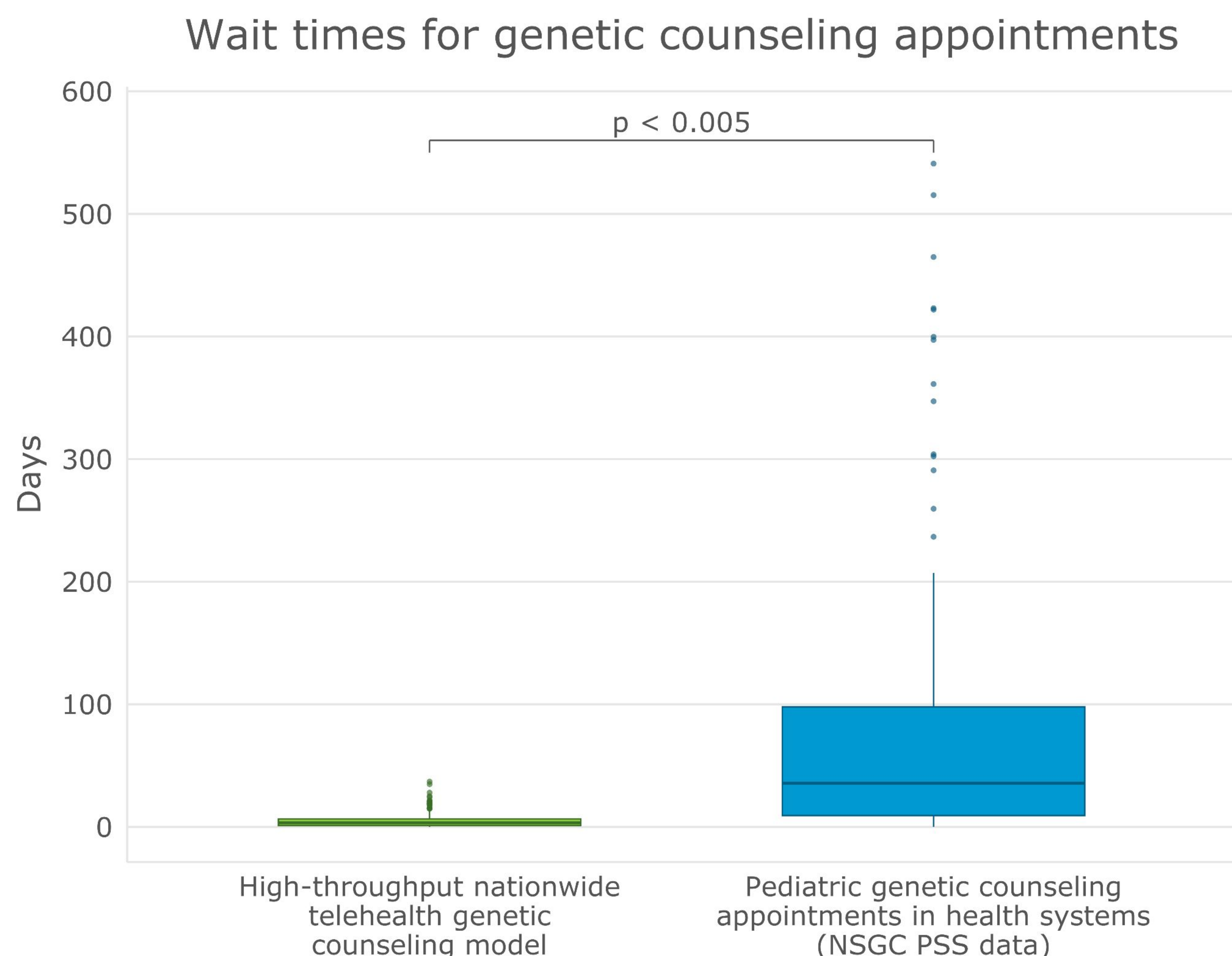


Pediatric patients with disease-causing genotypes in gNBS genes* seen for post-test genetic counseling at Genome Medical.



Time from scheduling to visit was compared to nationwide data from the NSGC Professional Status Survey on wait-times for pediatric genetic counseling in health systems (without MD)

High-throughput nationwide telehealth genetic counseling can provide timely gNBS results disclosure



Service Delivery Model

- Telehealth serving the US & Canada
- 28-40 patients per GC per week
- High throughput supported with technology, GCAs, practice efficiencies

Results

n	Median Age	Sex
377 patients	4.9 years (IQR 8.7 yr)	43.8% female (165/377)

Patients from **42** states.
Most frequently:



- Texas (13.5%)
- Florida (13.0%)
- California (9.0%)
- Georgia (6.9%)
- New York (4.5%)

Disease-causing genotypes in **123/667** gNBS genes.
Most frequently:



- *G6PD* (8.0%)
- *DMD* (5.6%)
- *SCN1A* (5.0%)
- *KCNQ1* (4.2%)
- *CFTR* (2.9%)

Patients seen within
3 days of scheduling

43.7% Study Cohort
7.0% National Avg. | $p < 0.005$

Median appointment duration
29 min (IQR 16 min)

Conclusions

- Markedly shorter wait-times vs similar appointments in health systems.
- Short wait-times may drive telehealth use even when local alternatives exist.
- High-throughput nationwide telehealth models may help meet the results return need of expanded gNBS.

Meeting the Genetic Counseling Demand for Genomic Newborn Screening Results:
A Nationwide Telehealth Service Delivery Analysis

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