



Expanding Access to Genomic Testing

Why Medicaid coverage matters in rare disease diagnosis



Our mission is to deliver personalized and actionable health insights to inform diagnosis, direct treatment, and improve drug discovery.



CASE STUDY

Years of searching.
One test provided answers in weeks.

Meet Amelia[†]

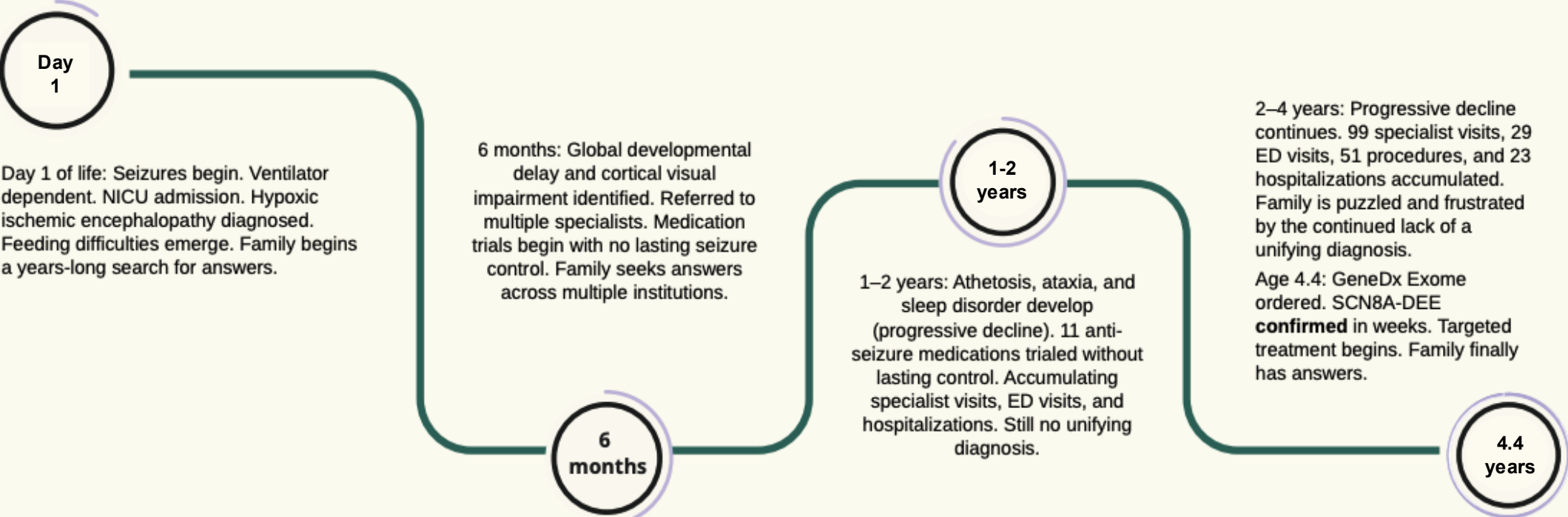
From day 1 of life, she experienced unrelenting seizures that required NICU ventilator support. Over the next four years, her family navigated specialist visits, emergency hospitalizations, and eleven medication trials without a unifying diagnosis. One exome test changed everything.



[†] Photo does not represent actual patient.



Her diagnostic journey took 4.4 years. It didn't have to.



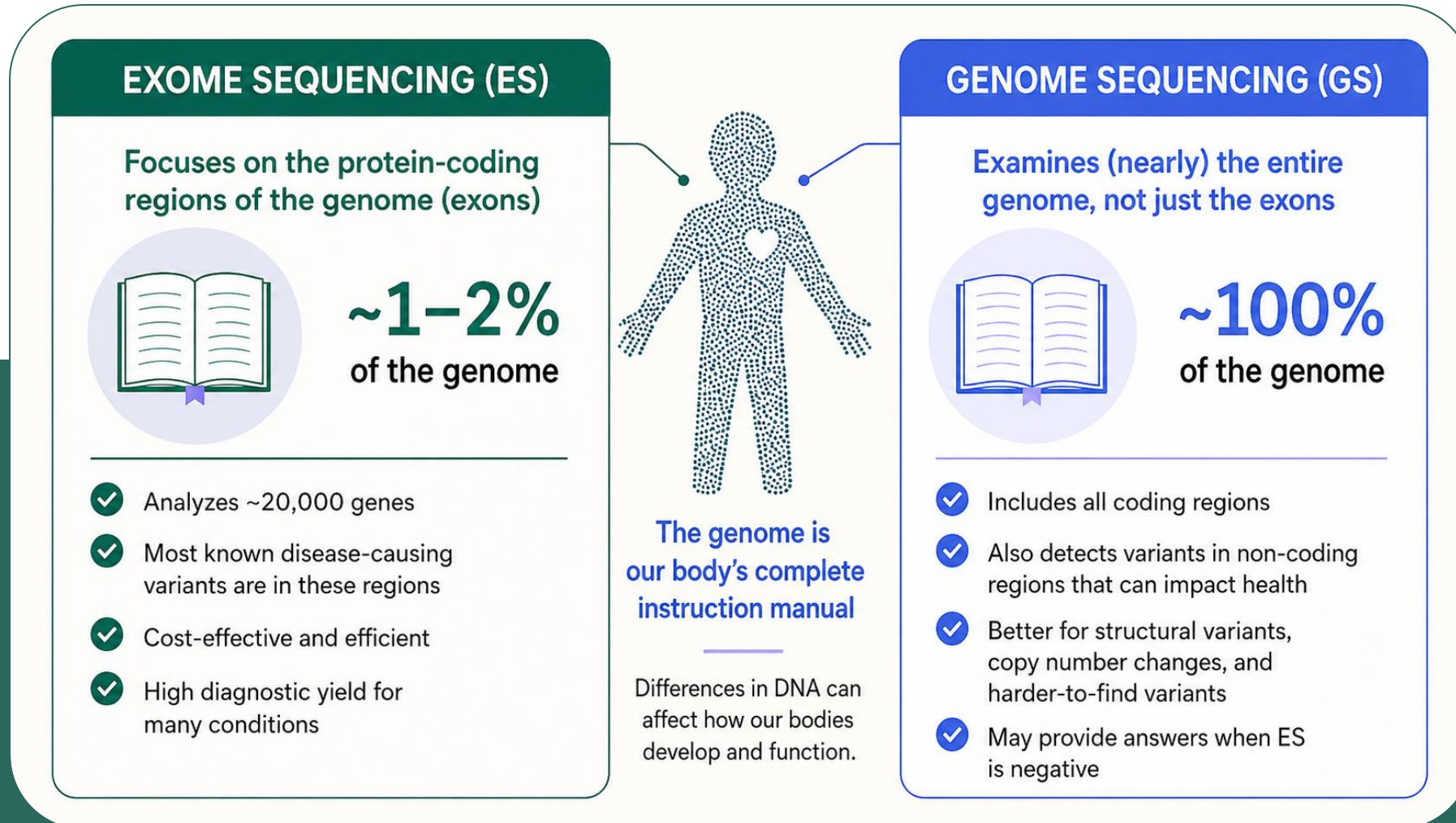
HEALTHCARE UTILIZATION

99 Specialist Visits	29 ED Visits
51 Procedures	23 Hospitalizations

POST-DIAGNOSIS REDUCTIONS

▼ 65% Specialist Visits	▼ 96% ED Visits
▼ 98% Procedures	▼ 98% Hospitalizations

Exome and genome sequencing are powerful diagnostic tools for complex unexplained conditions



Patients who benefit are already in your Medicaid program

Nearly half of children with special healthcare needs are covered by Medicaid

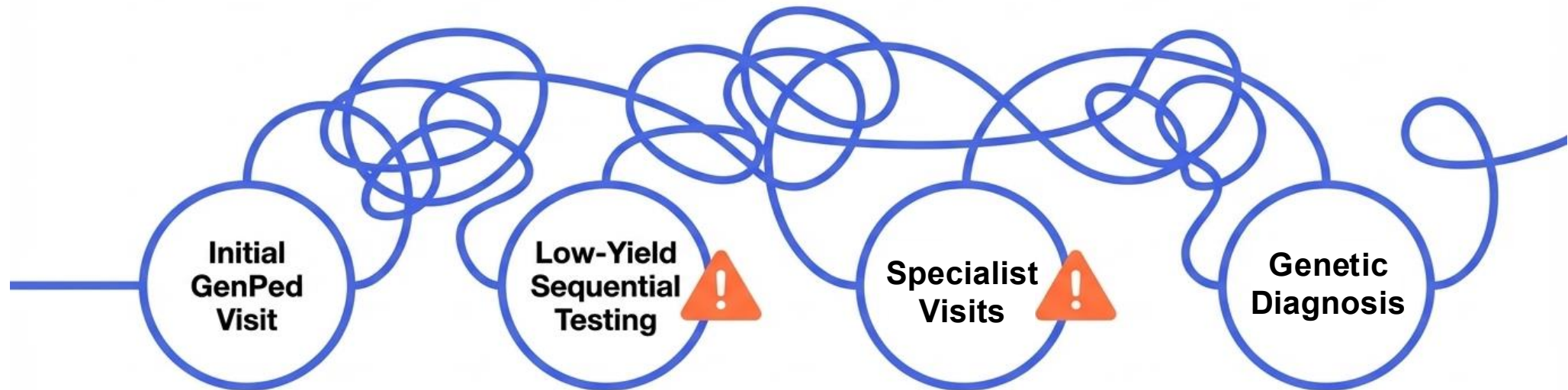
Patients typically have 1 or more of the indications below:

- Congenital abnormalities (birth defects)
- Significant Intellectual disability
- Global developmental delay
- Seizures/epilepsy
- Failure to thrive or other growth concerns
- Autism spectrum disorder
- Complex neurodevelopmental disorder
- Severe neuropsychiatric condition
- Cerebral palsy
- Dysmorphic features
- Significant hearing or visual impairment
- Period of unexplained developmental regression
- Biochemical findings suggesting inborn error of metabolism
- Family history strongly suggestive of a genetic etiology



The diagnostic odyssey is a winding search for answers

On average, a patient and their family experiences:



An exome or genome approach is recommended by experts

For patients with developmental delay, intellectual disability, congenital anomalies, epilepsy, cerebral palsy, or autism spectrum disorder, exome and genome are supported as first-line tests.¹⁻⁴

American College of Medical Genetics and Genomics (ACMG) recommends exome and genome as first-line tests

For patients with one or more congenital anomalies <1 year of age or for patients with developmental delay/intellectual disability with onset before age 18 (July 2021)²



National Society of Genetic Counselors (NSGC) recommends exome and genome as first-line tests

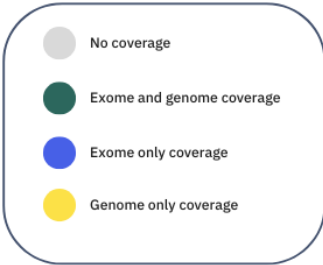
For all individuals with unexplained epilepsy, regardless of age or the presence of comorbidities. This recommendation is **endorsed by American Epilepsy Society (AES)** (Sept 2022)³



The American Academy of Pediatrics (AAP) recommends exome and genome as first-line tests

For children with global developmental delay (GDD) and intellectual disability (June 2025)⁵





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Medicaid coverage has grown, but disparities still exists compared to commercial insurance plans



Genome sequencing



Exome sequencing



Medicaid



Commercial

Nearly half of children with special healthcare needs are covered by Medicaid or CHIP

44%

of children with special health care needs rely on Medicaid or CHIP.



High Clinical Need

Medicaid disproportionately insures children with disabilities, complex conditions, and rare disease presentations.



No Self-Pay Alternative

Families cannot bridge a coverage gap out of pocket. When Medicaid does not cover testing, many children are left without answers.



EPSDT Obligation

Federal law requires Medicaid to cover medically necessary services for members under 21, including diagnostic testing.

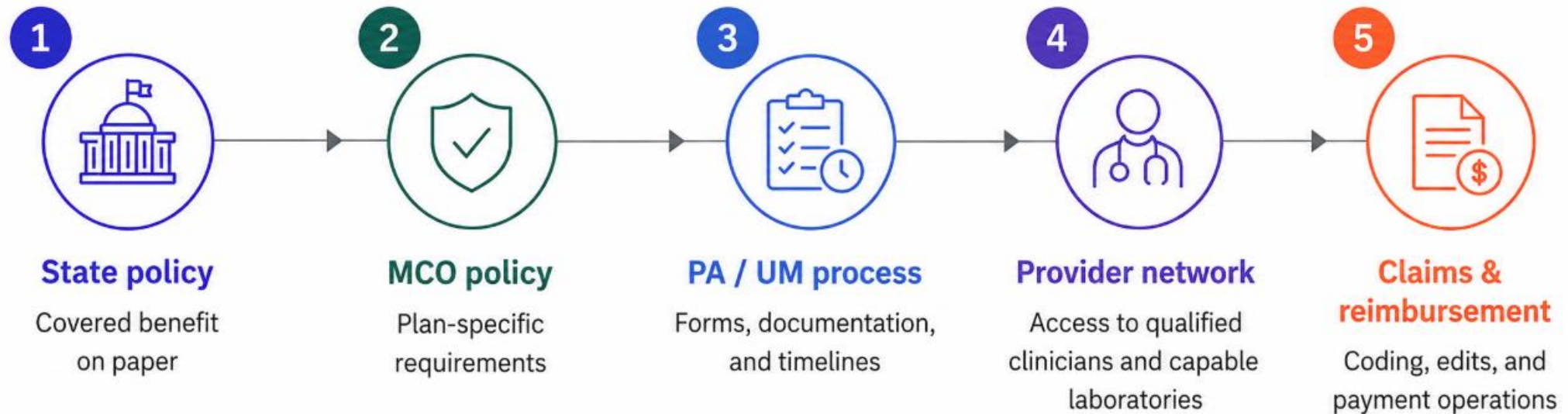


Geographic Equity

Access to a diagnosis should not depend on where a child lives or whether the family has private insurance.

A state can intend to cover genomic testing and still fail to deliver access

Coverage can break down at multiple points between policy and the patient



Real-world evidence shows access to ES/GS leads to significant cost savings

EPILEPSY COHORT

61% decrease in total healthcare costs



\$130,048 PPPY

Average cost before ES/GS



\$50,798 PPPY

Average cost 12 months after ES/GS

DD/ID[†] COHORT

Significant drop in acute care utilization



↓79%

Hospitalizations



↓48%

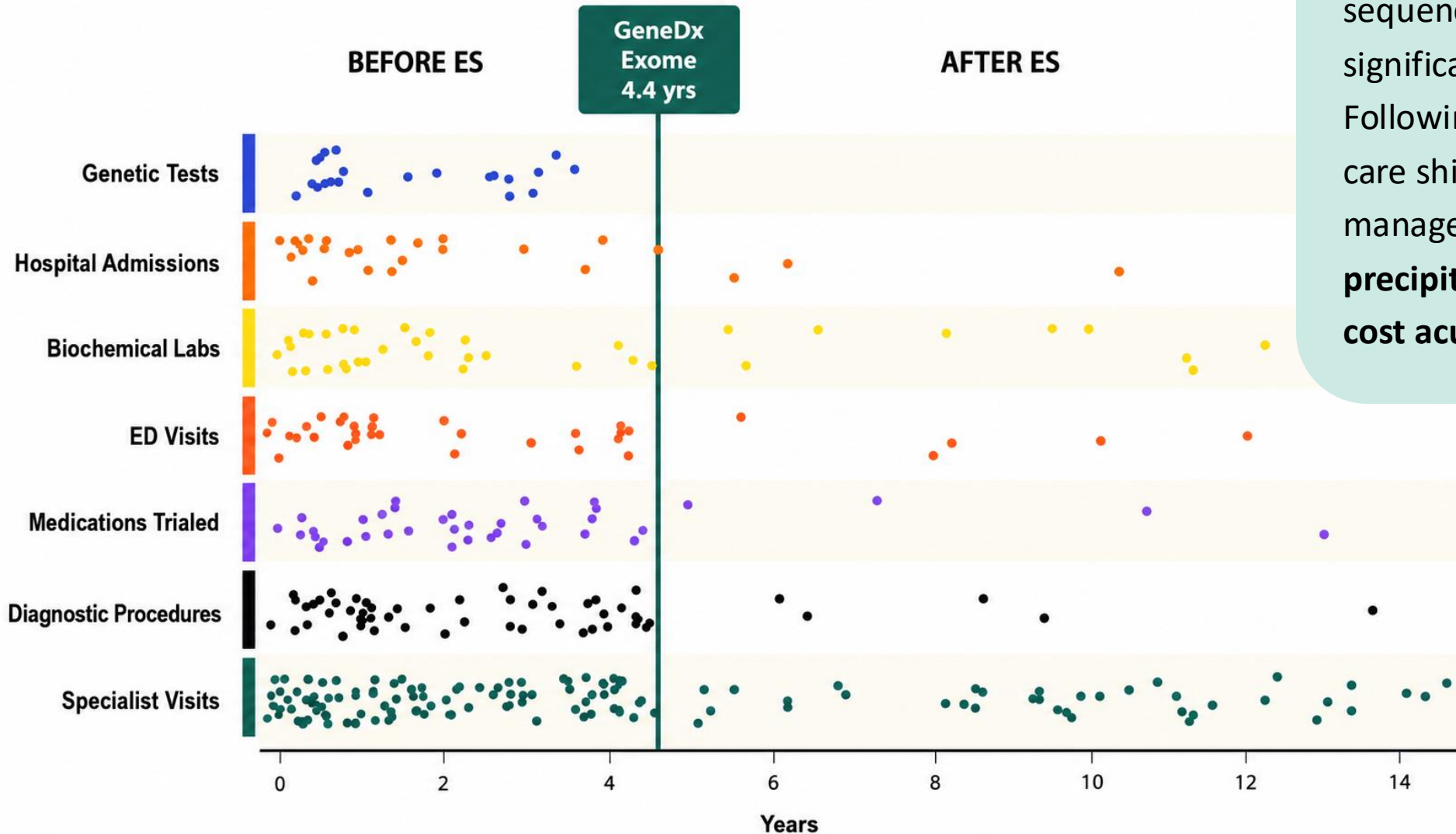
Emergency Dept visits

Healthcare resource utilization shifts from reactive to proactive

[†] DD/ID: Developmental Delay / Intellectual Disability

Amelia's healthcare utilization timeline

Individual events plotted from symptom onset through end of follow-up



Key Finding: The delayed application of exome sequencing (ES) represents significant "diagnostic waste." Following ES, the trajectory of care shifted to precision management, resulting in a **precipitous decline in high-cost acute care utilization.**

Three solvable barriers are blocking access

These gaps aren't inevitable. They result from policy choices that can be changed.

1 Policy



Medicaid Playing Catch-Up

Medicaid coverage has grown but still lags behind commercial insurance—despite guidelines and EPSDT obligations.

2 Provider Access



Specialist Shortage + Specialist Requirements

Many policies require specialist ordering in a workforce too small to meet demand—a self-defeating barrier.

3 Administrative Friction



Unrealistic UM Requirements

Variability in prior auth requirements across plans leads to frequent denials unrelated to medical necessity.



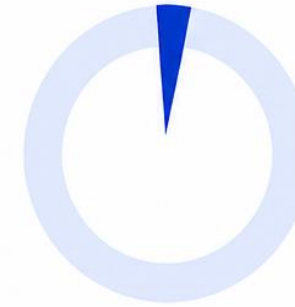
Each barrier carries a real cost—to the family waiting years for a diagnosis, and to the payor funding that wait.

Demand is growing, yet genetics specialists are in short supply



2/500,000

There are only **2 medical geneticists** for **every 500,000 people** in the United States.



<1%

Medical geneticists account for less than 1% of the total physician workforce.



53.4%

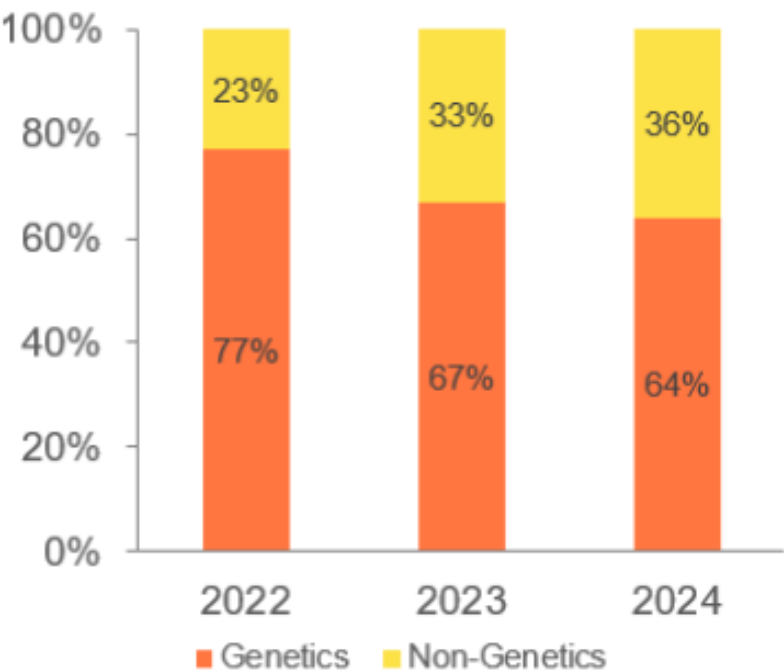
Primary Care Physicians report having no access to genetics expertise.



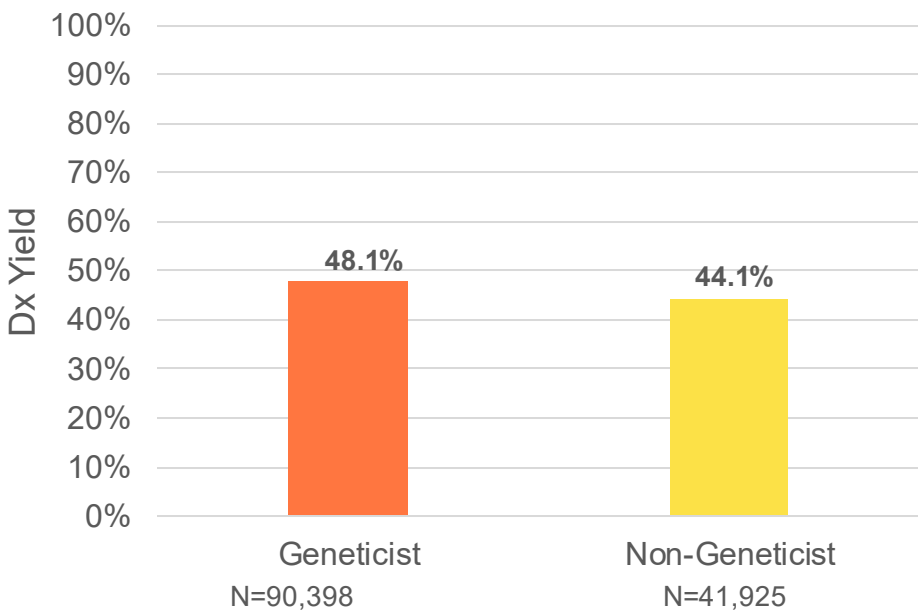
The shortage leads to lengthy wait times for specialist evaluations and significant travel burdens for patients and families, delaying optimal care.

Real-world evidence shows non-genetics providers identify appropriate patients for genomic testing

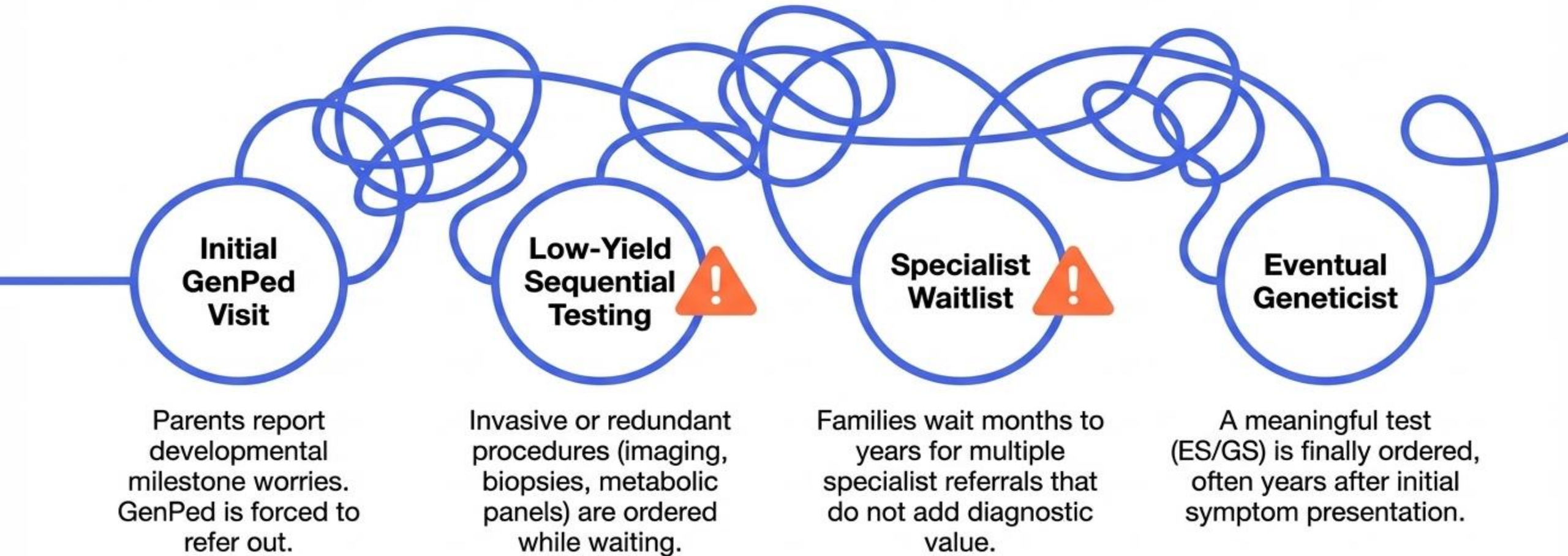
Non-genetics providers now order more than one-third of ES/GS tests



Diagnostic yield is nearly identical across ordering provider type



Lack of access creates unnecessary delays and drives higher costs through long and winding diagnostic odysseys



Nevada Payor Coverage Landscape

Segment	Total Lives	Contracted %	GS %	ES %	ES or GS %	rGS %
Commercial	2.5M	15.9%	94.1%	95.7%	95.7%	89.4%
Medicaid (FFS + Managed)	684,967	7.7%	0.0%	0.0%	0.0%	0.0%
Tricare / Military	107,291	0.0%	0.0%	0.0%	0.0%	0.0%*

* Tricare rGS status not confirmed in tracker.



NEVADA LAW REQUIRES COVERAGE FOR BIOMARKER TESTING



Nevada Revised Statutes (NRS) 422.272364 (Medicaid) and 695G.1703 (Managed Care) require coverage of medically necessary biomarker testing for diagnosis, treatment, management and monitoring of cancer when supported by:

- FDA-labeled indications
- National/Local Coverage Determinations (42 C.F.R. § 400.202)
- Nationally recognized clinical practice guidelines
- Or other medical & scientific evidence



EPSDT REQUIRES COVERAGE OF ALL MEDICALLY NECESSARY SERVICES



Medicaid must cover all services that correct or ameliorate defects and physical/mental illnesses and conditions discovered by screening, consistent with EPSDT.⁴

Restrictive policies (e.g., step therapy, specialty requirements) conflict with EPSDT.

Managed Medicaid	Anthem	188,114
	Caresource Nevada	88,177
	Health Plan of Nevada	152,908
	Molina Healthcare	80,090
	SilverSummit Healthplan	117,521
Medicaid FFS	Nevada Medicaid (FFS)	52,669

Become the catalyst that ends the diagnostic odyssey

As Rare Disease leaders you can:

- ✓ Advocate for exome and genome sequencing as an essential diagnostic benefits under EPSDT
- ✓ Ensure aligned FFS and MCO implementation
- ✓ Advocate for earlier diagnosis by empowering non-specialists
- ✓ Advise for removal of administrative barriers that delay medically necessary testing
- ✓ Measure success by how quickly eligible children receive answers



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