



# One test. Big picture. Brighter futures.

GeneDx (Nasdaq: WGS)

April 29, 2024

GeneDx

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# WGS 1Q 2024 Results



First quarter 2024 revenue from continuing operations<sup>1</sup> of \$61.5M with 96% year-over-year revenue growth for exome and genome test revenue

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First quarter 2024 adjusted gross margins<sup>2</sup> for continuing operations of 61%

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Eight consecutive quarter of cash burn decline: first quarter 2024 was \$17M  
Ending March 31, 2024 cash, cash equivalents, marketable securities and restricted cash position of \$113.9 million

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Reiterating the path to profitability in 2025

<sup>1</sup> Results from continuing operations include exclude the results of the exited Legacy Sema4 diagnostic testing business. Total company results include GeneDx's continuing operations and the financial impacts of exited Legacy Sema4 business activities.

<sup>2</sup> Adjusted gross margin is a non-GAAP financial measure. A reconciliation of GAAP and non-GAAP results is provided in the Company's earnings release dated April 29, 2024.

## Updating 2024 Guidance

Drive full year 2024 revenues between \$235 to \$245 million  
(previously \$220-230 million)

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Expand full year 2024 adjusted gross margin profile to at least  
60% (previously at least 50%)

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Use \$70 to \$80 million of net cash for full year 2024  
(previously \$75-80 million)

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Turn profitable in 2025 (no change)

# Revenue - strong growth driven by high value whole exome and genome

**91%**

Increase in 1Q24 exome/genome test result volume year-over-year; +6% sequentially

**\$61.5M**

First quarter 2024 revenue from continuing operations<sup>1</sup>

Revenue <sup>1</sup>	Q1 2024
Revenue from continuing operations	\$61.5M
<i>Growth year-over-year</i>	<b>51%</b>
<i>Growth sequentially</i>	<b>6%</b>
Exome and genome test revenue	\$44.0M
<i>Growth year-over-year</i>	<b>96%</b>
<i>Growth sequentially</i>	<b>12%</b>

<sup>1</sup> Total company revenues were \$62.4M for the first quarter 2024. Results from continuing operations exclude the results of the exited Legacy Sema4 diagnostic testing business. Total company results include GeneDx's continuing operations and the financial impacts of exited Legacy Sema4 business activities.

# Gross profit – expansion driven by mix shift, cost per test reductions

Exome/genome can be the best test for patients. They are also best for our business.

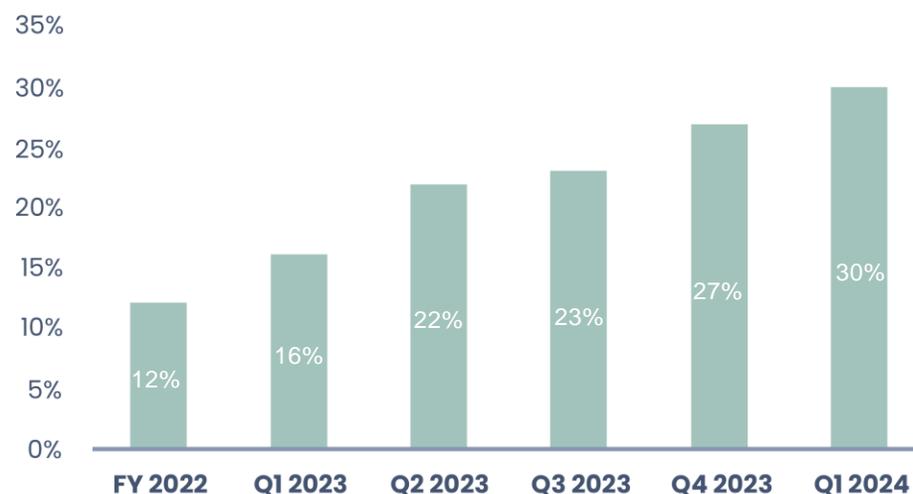
30%

Exome/genome test result volume

61%

Adjusted gross margins<sup>1</sup> for continuing operations in 1Q24, up from 56% in Q4 2023 and 34% in Q1 2023

**Exome/Genome Test Mix**



● Exome/Genome as a % of total test results

Adj. Gross Profit for continuing operations	1Q24	QoQ Sequential	YoY
Adj. Gross Profit <sup>1</sup>	\$37.4M	+15%	+169%
Adj. Gross Margin % <sup>1</sup>	61%	+489bps	+2,671bps

## Cash - Balance sheet bolstered to execute growth strategy

**\$114M<sup>1</sup>**

Cash, cash equivalents, marketable securities and restricted cash on hand March 31, 2024

**\$17M<sup>2</sup>**

Net use of cash for the total company in Q1

**71%**

Improvement in total company net cash burn rate year-over-year; improved 48% sequentially

**8**

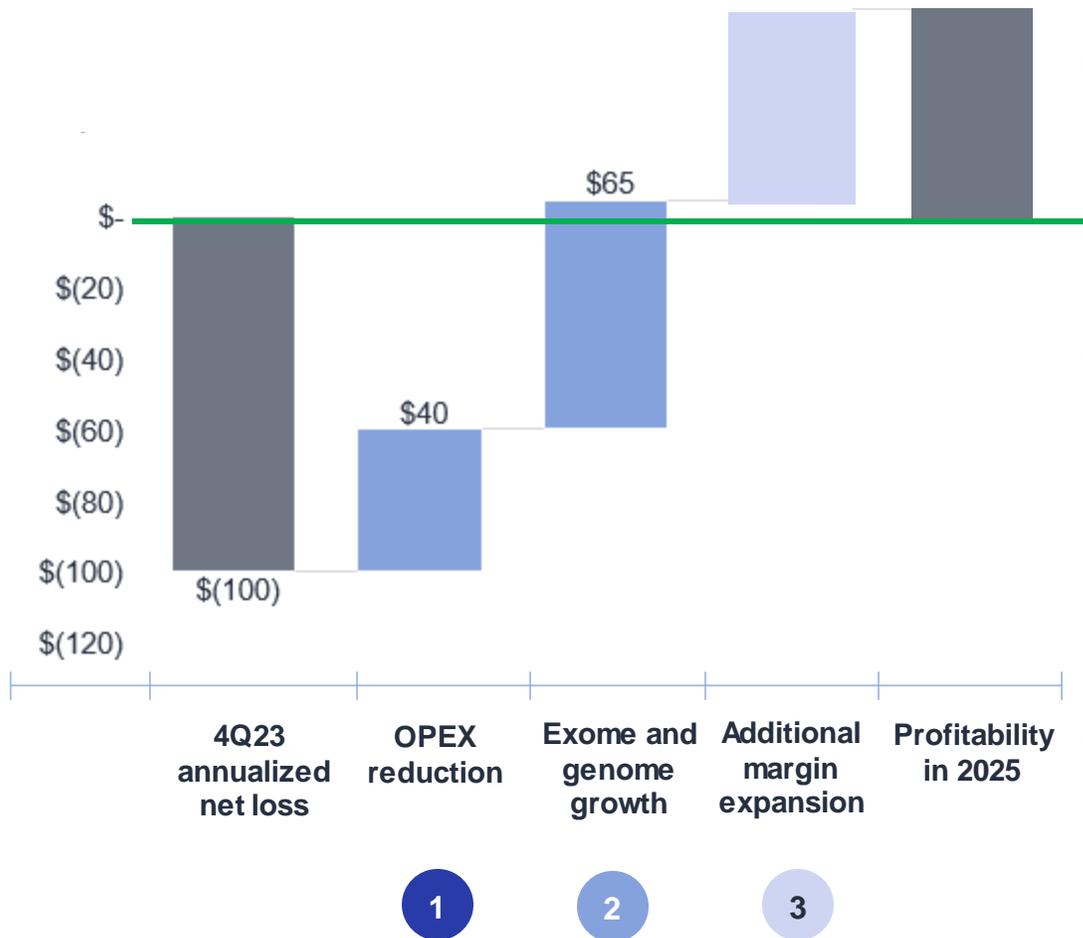
Consecutive quarters of cash burn reduction since acquiring GeneDx

<sup>1</sup> An additional \$25M in debt capacity is contingently available to use under the five-year senior secured credit facility with Perceptive Advisors entered into on October 27, 2023. The agreement provides access of up to \$75 million, consisting of an initial tranche of \$50 million, which has been drawn, and a subsequent tranche of \$25 million is available – at the Company's option, subject to certain timelines and other defined criteria.

<sup>2</sup> First quarter 2024 net cash burn included approximately \$6 million to fund the company's annual 401K employer match, approximately \$2.9 million in one-time payments to pay down previously reserved Legacy Sema4 refund requests and \$800 thousand in severance payments related to our previously announced cost reduction initiative.

# Our path to profitability and beyond

## What we need for profitability and beyond



## How we will achieve those goals

### 1 OPEX reduction plan

- On October 30, 2023, we enacted a plan to remove approximately \$40M in annual operating expense including no less than \$20M through headcount reductions which have already been completed

### 2 Exome and genome growth

- Revenue growth through 2025 expected to be driven by whole exome/genome testing, which operates at >60% gross margins
- Assuming no change in the current margin profile, increasing exome/genome's annual gross profit by \$65M requires us to maintain growth rates in 2024 and 2025 at nearly half of what was achieved in 2023
- As exome/WGS takes more share of test mix from low/negative margin legacy panels, gross margin will rise

### 3 Additional opportunities

- Beyond break-even, we see significant additional upside in –
  - Further reducing exome/WGS COGS as we benefit from scale and certain in-flight AI/automation initiatives
  - Improving collection rates across the portfolio as payor coverage expands and we increase internal performance regarding collection/reimbursement performance
  - Acceleration of our biopharma/data business
  - Untapped long-term opportunities providing WGS newborn screening, interpretation as a service, and others

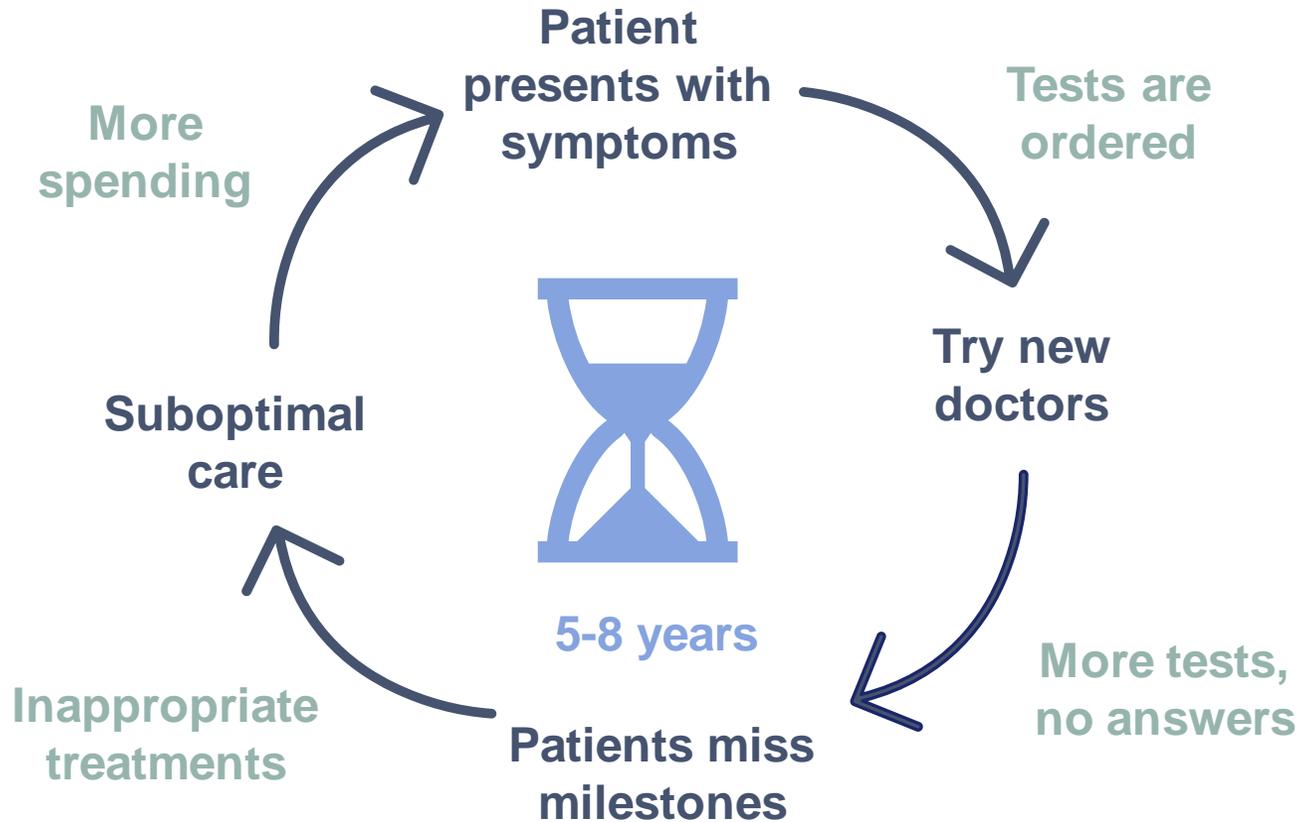
**One test.  
Big picture.  
Brighter futures.**



# Appendix

**GeneDx is a leader in improving health outcomes through genomic insights.**

# The problem: Lack of genomic data can lead to a cycle of misdiagnosis and suboptimal care



## Missed diagnoses impact everyone

### Patients/Caregivers

- Inappropriate treatments and delayed care
- Suffering

### Healthcare systems

- Increased costs and burden
- Missed opportunities to develop treatments

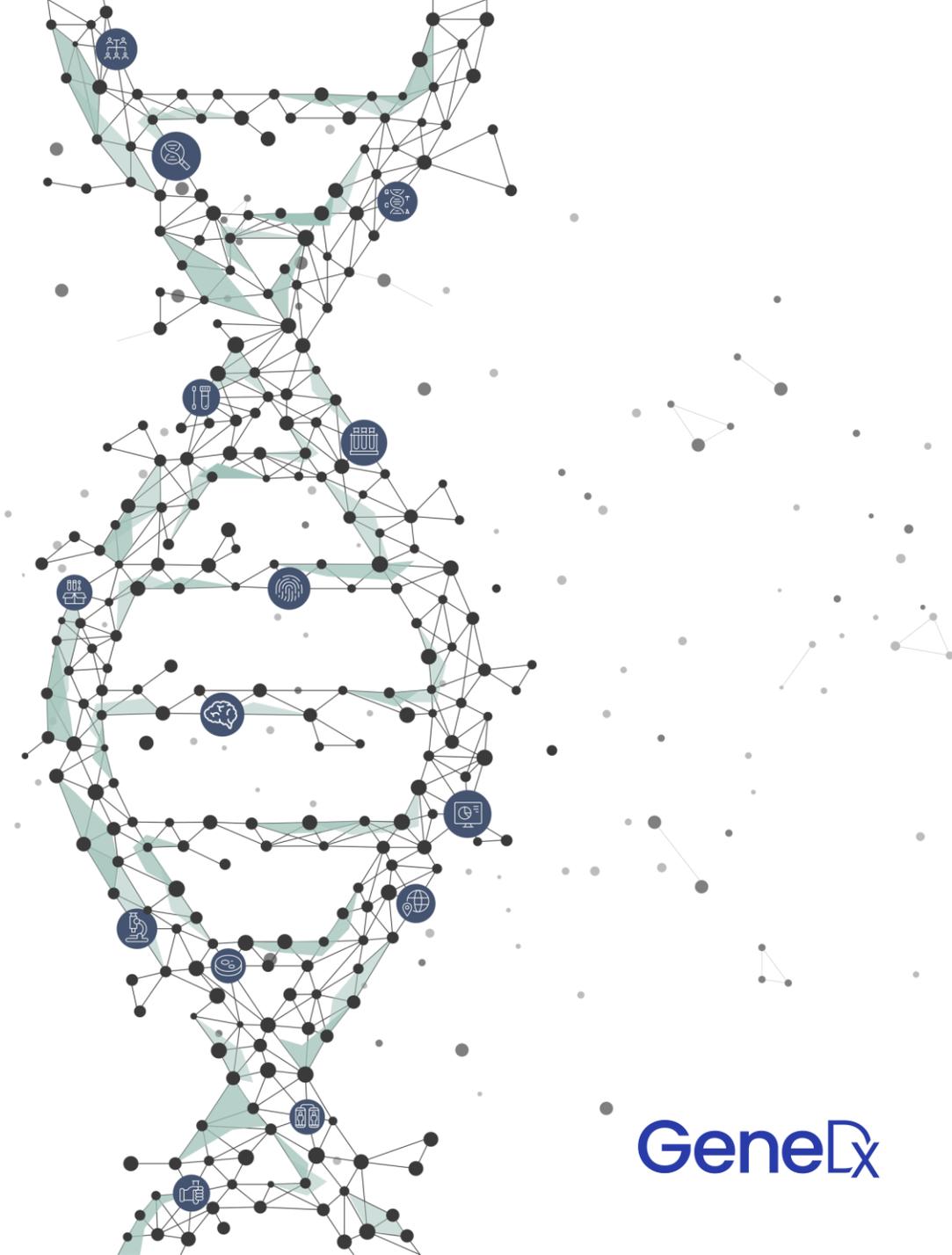
### Providers

- Stress from continued patient suffering
- Inconclusive diagnoses

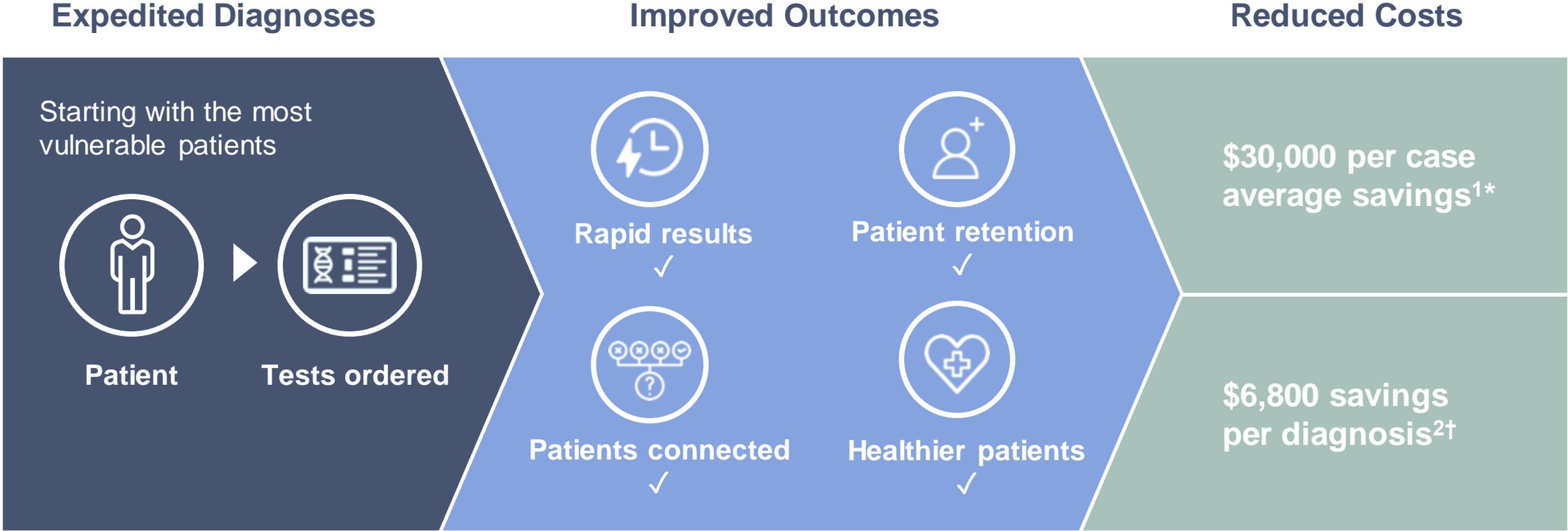
# GeneDx offers leading exome and genome products

*Translating complex genomic data into definitive diagnoses for patients*

- **Genome sequencing** - Analyzes the entirety of an individual's DNA, which is known as the genome. The genome includes ~20,000 genes.
- **Exome sequencing** – Analyzes the protein coding regions of the ~20,000 genes in an individual's genome, which is known as the exome. The exome is thought to contain a majority of disease-causing genetic variants.



# Exome sequencing can break the cycle of misdiagnosis and uncertainty



\*In the NICU from reduced length of stay, unnecessary care (inpatient).  
†When tested at first tertiary presentation for Pediatric Delay Disorder (outpatient).

**References:** 1. ScienceDaily. (2017, October 19). Rapid whole-genome sequencing of neonatal ICU patients is useful and cost-effective. ScienceDaily. 2. Tan TY, Dillon OJ, Stark Z, et al. Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. *JAMA Pediatrics*. 2017;171(9):855. doi:10.1001/jamapediatrics.2017.1755



# Changing the perception of exome and genome sequencing

GeneDx has spent over a decade solving for limitations of the past and differentiating our products

Then

GeneDx Now



Turnaround time

“These tests take months to get results”



“I can get results in days to weeks”



Cost

“These tests are wildly expensive”



“Tests are affordable and widely covered”



Interpretation

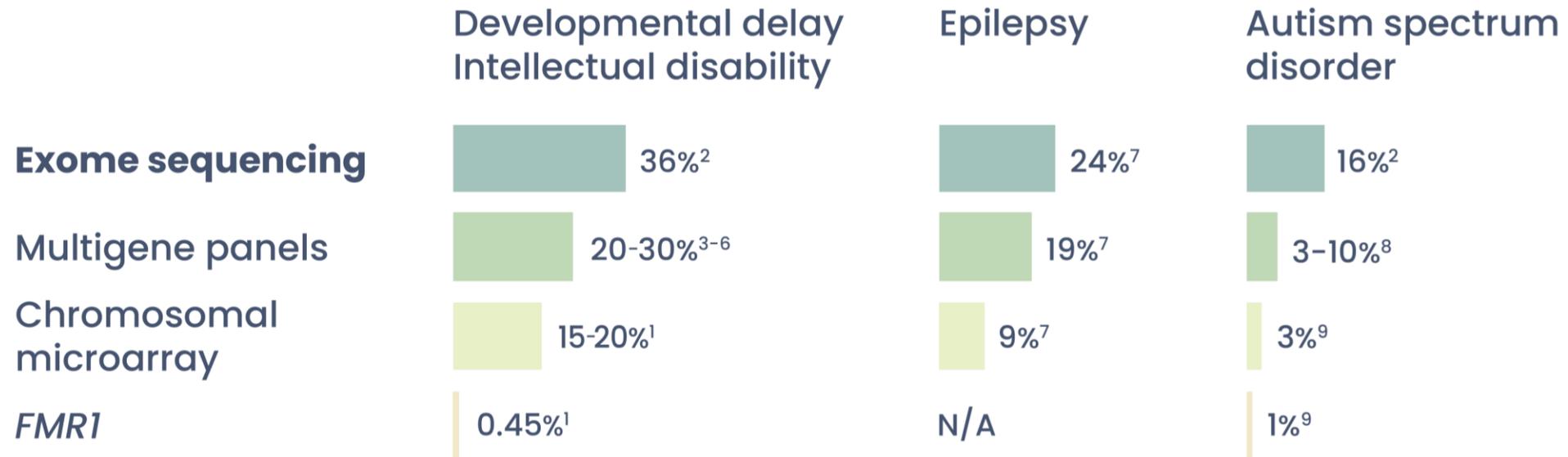
“Results are confusing and filled with useless information”



“My patient can get a definitive diagnosis”

GeneDx

# Exome sequencing offers greater diagnostic yields vs. other technologies



1. Savatt JM *et al. Front Pediatr.* 2021;9:526779. 2. Srivastava S *et al. Genet Med.* 2019;21(11):2413–2421. 3. Pেকেles H *et al. Pediatr Neurol.* 2019;92:32-36. 4. Stefanski A *et al. Epilepsia.* 2021;62(1):143-151. 5. Mellone S *et al. Front Genet.* 2022;13:875182. 6. Spataro N *et al. Genes (Basel).* 2023;14(3):708. 7. Sheidley BR *et al. Epilepsia.* 2022;63(2):375-387. 8. Ni Ghráiligh F *et al. J Autism Dev Disord.* 2023;53(1):484-488. 9. Arteché-López A *et al. Genes.* 2021(12):560.

**GeneDx is positioned to enable a data-informed future for healthcare.**

# New market expansion enables us to serve more patients

GeneDx is starting with a focus on rare disease and pediatrics and then expanding into larger markets



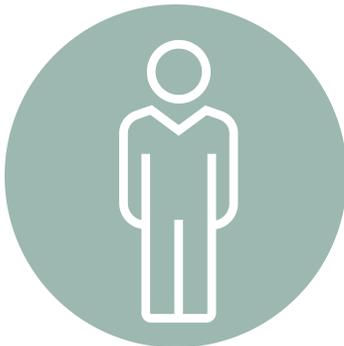
**Rare Disease  
& Pediatrics: \$3B**

Rapidly growing patient opportunity and substantial cost savings via early screening



**Newborn  
Screening: \$10B**

Currently participating in studies to evaluate exome and genome sequencing at birth



**Adults: \$16B**

Expanding into adult markets to replace multi-gene panel and individual gene tests

Conservatively, our total addressable market is **~\$30 billion.\***

\*Company Estimates supported by DefinitiveHC diagnosis data.

# We're focused on the Rare Disease & Pediatrics market today



## Rare Disease & Pediatrics: \$3B

Rapidly growing patient opportunity and substantial cost savings via early screening

### Inpatient

#### Target Clinicians:

- Geneticists
- Neonatologists

#### Products:

- Primarily rapid whole genome sequencing

#### Primary Clinical Indications:

- Unexplained critical illness
- Congenital anomalies

**Inpatient**  
~1/3 of TAM

**Outpatient**  
~2/3 of TAM

### Outpatient

#### Target Clinicians:

- Geneticists
- Pediatric Neurologists
- Developmental Pediatricians
- Pediatricians (long-term)

#### Products:

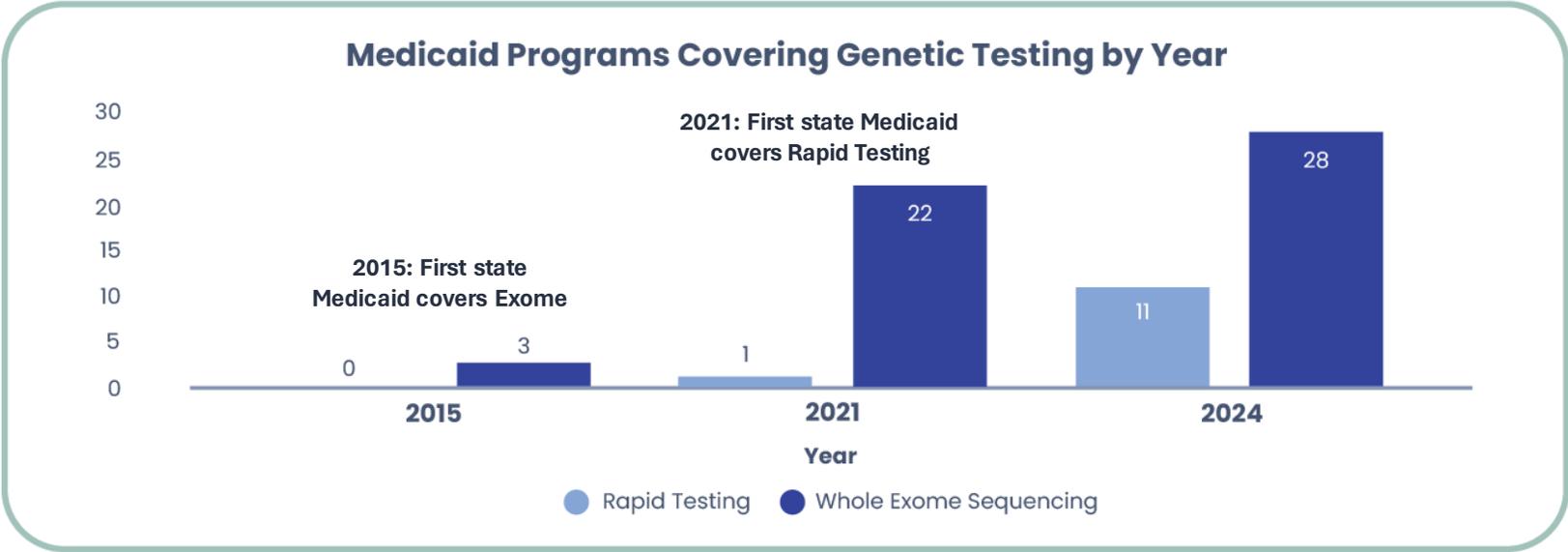
- Primarily exome and growing genome

#### Primary Clinical Indications:

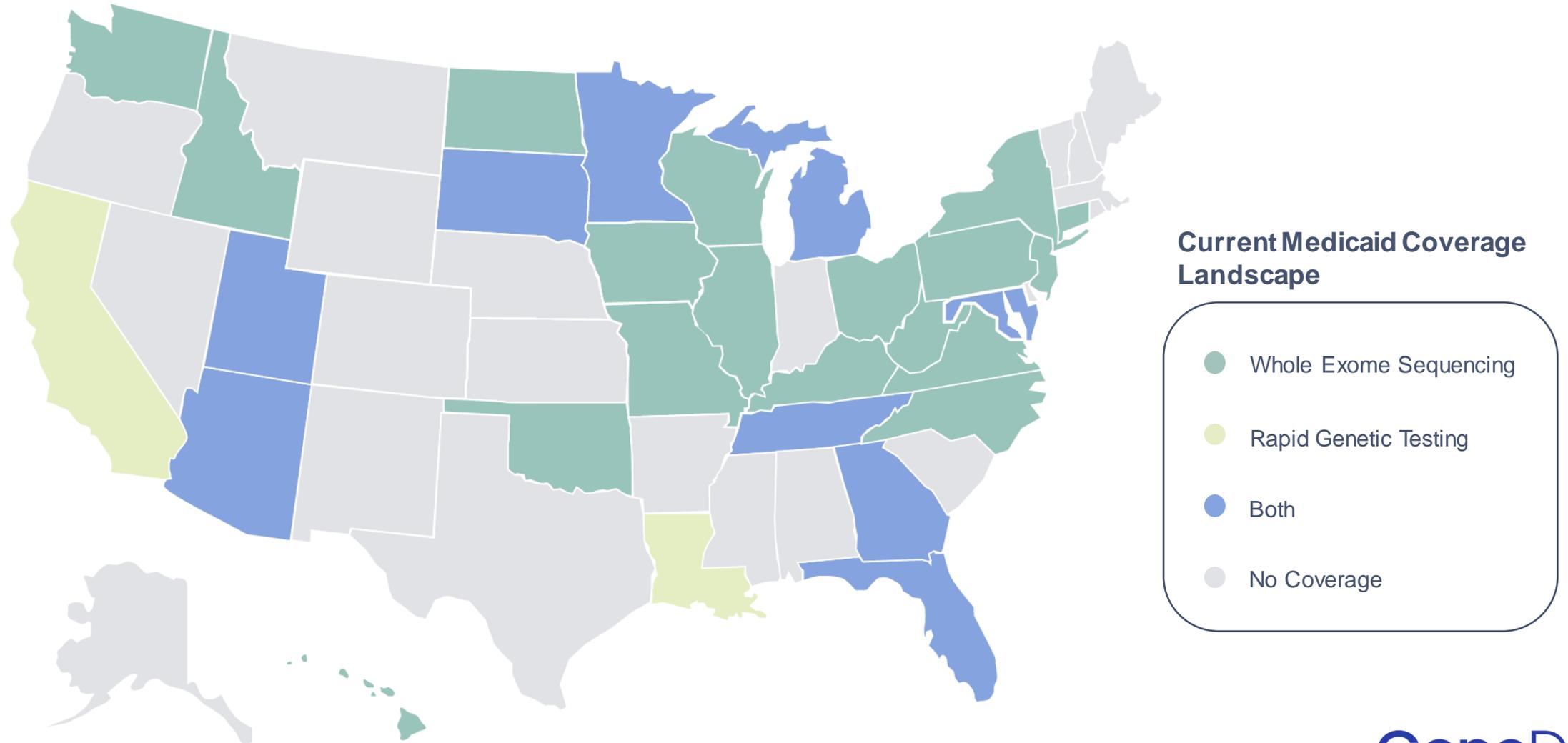
- Epilepsy
- Intellectual disability
- Developmental delay

# Payor coverage for exome and genome sequencing is expanding

- › GeneDx is **contracted with over 80% of covered lives**, including all large national commercial payers
- › **Medicaid and commercial insurance coverage continues to grow** for exome and genome
  - **28 states** cover exome sequencing
  - **11 states** cover rapid genome sequencing
  - **Biomarker bills** are driving momentum in Medicaid coverage in key states like Texas and California



# Medicaid programs across the country are expanding access



# Medical practice guidelines recommend exome and genome sequencing for patients



## ACMG Practice Guideline<sup>1</sup>:

*“Strong recommendation based on the available evidence to support the use of ES/GS as either a first- (or second-) line test in patients .... ES/ GS demonstrates clinical utility for the patients and their families with limited evidence for negative outcomes and the ever-increasing emerging evidence of therapeutic benefit.”*



## NSGC Guideline<sup>2</sup>:

*“Recommending Exome Sequencing as a First-Tier Genetic Test for Unexplained Epilepsies”*

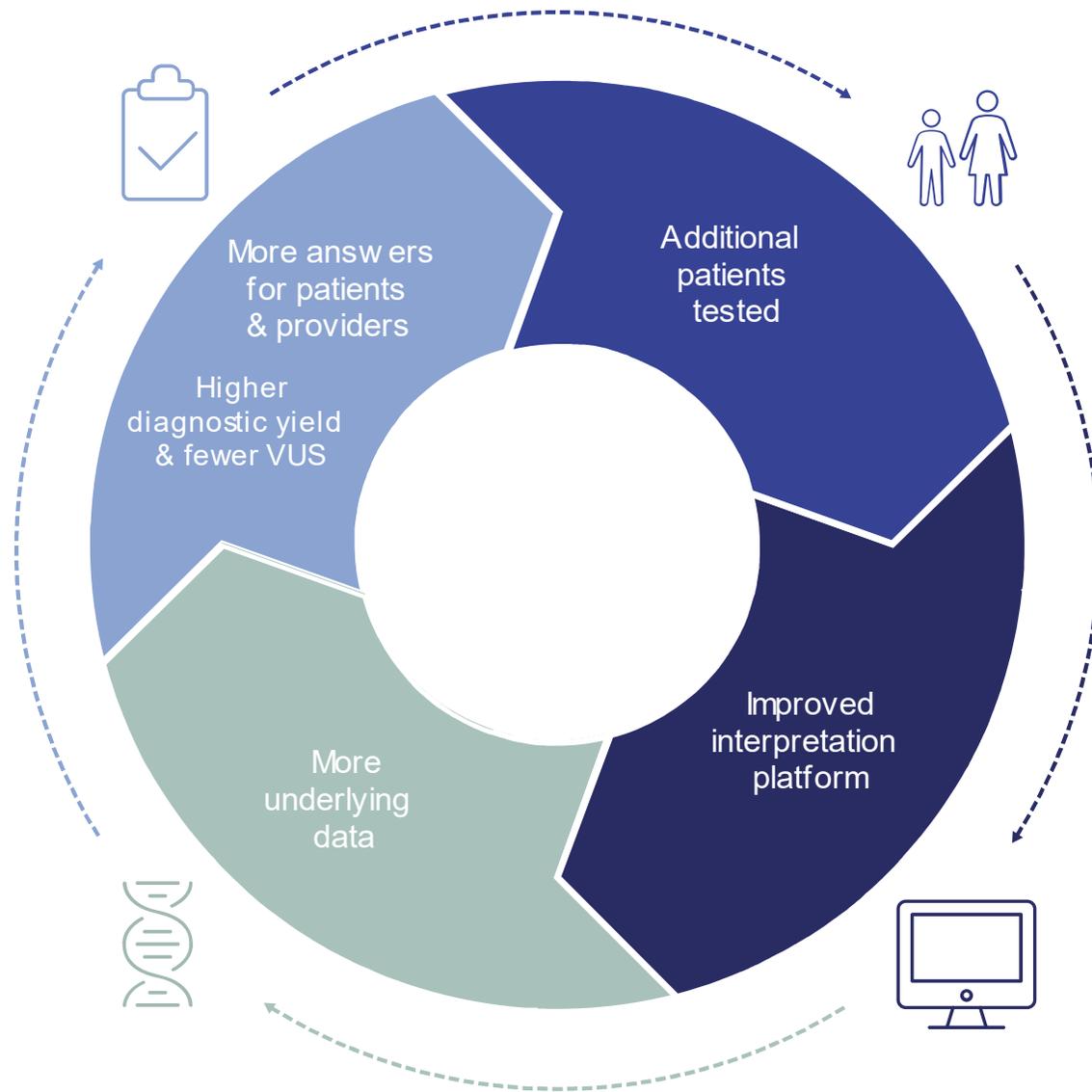


## American Epilepsy Society:

*“Exome or genome sequencing are favored for most scenarios, as they are more likely to provide a diagnosis.”*

1 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 Nov;23(11):2029-2037. doi: 10.1038/s41436-021-01242-6. Epub 2021 Jul 1.

2 Smith L, Malinowski J, Ceulemans S, et al. Genetic testing and counseling for the unexplained epilepsies: An evidence-based practice guideline of the National Society of Genetic Counselors. *J Genet Couns*. 2022 Oct 24. doi.org/10.1002/jgc4.1646



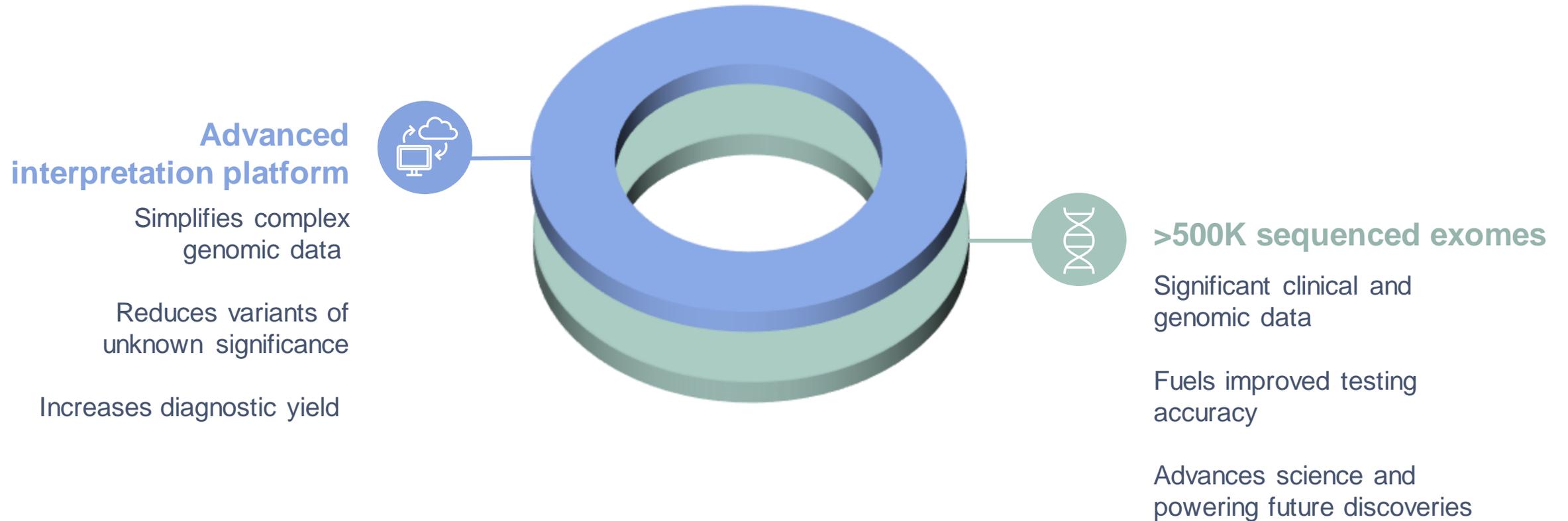
## Pay-it-forward data strategy

For every patient that we test, our underlying interpretation platform gets smarter, and we can offer more answers to more patients.

**The impact scales as we capture more and more of the market.**

# Data is at the center of our business

Our huge dataset and intelligent interpretation platform set us apart and fuel innovation



# Common diseases are in fact a constellation of genetic diagnoses

One example is epilepsy. At least 768 different genes are related to seizures.



Only 43% are tested on many commercial epilepsy panels

# Common diseases are in fact a constellation of genetic diagnoses

One example is epilepsy. At least 768 different genes are related to seizures.



Only 43% are tested on many commercial epilepsy panels



Exome sequencing checks all 768 genes

**Today, we shorten the diagnostic journey.  
Tomorrow, we hope to prevent it.**

# Building the future: NICU

Shorter hospital stays. Less uncertainty. Better care.

In phase one of the SeqFirst study, 125 infants were offered rapid WGS:



**63%** of infants had abnormal rapid WGS results, and **88%** of these cases resulted in a change in management



**1 in 4** infants with abnormal results were not previously suspected to have a genetic syndrome



Families of enrolled infants reported an **overall positive experience**, regardless of rapid WGS test outcome



# Building the future: Newborn screening

Fast diagnosis. Clear next steps.  
Reduced healthcare costs.



Screened **5,000** healthy infants, toward goal of >100,000



**4.3%** positive rate



Average age of diagnosis for these conditions is **7-11** years old

GeneDx



# Building the future: Partnerships

Enriched data. Empowered  
drug discovery. Improved outcomes.

GeneDx offers solutions across the  
pharma drug development pipeline

*Find*

*Connect*

*Understand*



# Building the future: Interpretation as a service

Scientific rigor. Medical value.  
Establishing genomics as the  
standard of care for all.

