

# GeneDx

## NASDAQ: WGS

*One Test: Miss Less. Discover More.*

October 30, 2023

The GeneDx logo is displayed in white text on a light blue rounded rectangular background. The 'D' in 'GeneDx' is stylized with a white outline and a blue fill.

## Disclaimer

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The forward-looking statements and opinions contained in this presentation are based on our management’s beliefs and assumptions and are based upon information currently available to our management as of the date of this presentation and, while we believe such information forms a reasonable basis for such statements, such information may be limited or incomplete, and our statements should not be read to indicate that we have conducted an exhaustive inquiry into, or review of, all potentially available relevant information. Many factors could cause actual future events to differ materially from the forward-looking statements in this presentation, including but not limited to: (i) the ability to implement business plans, goals and forecasts, and identify and realize additional opportunities, (ii) the risk of downturns and a changing regulatory landscape in the highly competitive healthcare industry, (iii) the size and growth of the market in which we operate, and (iv) our ability to pursue our new strategic direction. The information, opinions and forward-looking statements contained in this announcement speak only as of its date and are subject to change without notice.

## Use of Non-GAAP Financial Measures

This presentation includes non-GAAP financial measures, such as Adjusted Gross Profit (Loss), Adjusted Gross Margin and Adjusted EBITDA. We define Adjusted Gross Profit (Loss) as revenue less cost of services, excluding stock-based compensation expense and restructuring costs, and we define Adjusted Gross Margin as our Adjusted Gross Profit (Loss) divided by our revenue. We define Adjusted EBITDA as our net loss adjusted for interest expense, net, income tax expense, depreciation and amortization, stock-based compensation expense, transaction, acquisition and business integration costs, restructuring costs, change in fair market value of financial liabilities and other income. Management believes that these non-GAAP measures of financial results are useful in evaluating the GeneDx's operating performance compared to that of other companies in its industry, as these metrics generally eliminate the effects of certain items that may vary from company to company for reasons unrelated to overall operating performance. Please refer to our earnings release for a reconciliation of GAAP to non-GAAP financial measures.

This presentation contains estimates, projections and other information concerning our industry, our business, and the markets for our products and services. Information that is based on estimates, forecasts, projections, market research or similar methodologies is inherently subject to uncertainties, and actual events or circumstances may differ materially from events and circumstances that are assumed in this information. Unless otherwise expressly stated, we obtained this industry, business, market and other data from our own internal estimates and research as well as from reports, research surveys, studies and similar data prepared by market research firms and other third parties, industry, medical and general publications, government data and similar sources. While we believe our internal company research as to such matters is reliable and the market definitions are appropriate, neither such research nor these definitions have been verified by any independent source.

We discuss these and other risks and uncertainties in greater detail in the sections entitled “Risk Factors” and “Management's Discussion and Analysis of Financial Condition and Results of Operations” in our periodic reports and other filings we make with the SEC from time to time. Given these uncertainties, you should not place undue reliance on the forward-looking statements. Moreover, we operate in a very competitive and rapidly changing environment. New risks emerge from time to time. Except as required by law, we undertake no obligation to update publicly any forward-looking statements for any reason after the date of this presentation to conform these statements to actual results or to changes in our expectations. We file reports, proxy statements, and other information with the SEC. Such reports, proxy statements, and other information concerning us are available [www.sec.gov](http://www.sec.gov). Requests for copies of such documents should be directed to our Investor Relations department at GeneDx Holdings Corp. 333 Ludlow Street, North Tower, Stamford, Connecticut, 06902. Our telephone number is 800-298-6470.

## Our Mission:

Deliver personalized and actionable health insights to inform diagnosis, direct treatment and improve drug discovery.



GeneDx

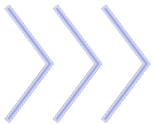
# 3Q 2023 and Recent Highlights



Grew whole exome and genome test volume by 71% year-over-year



Delivered total revenue of \$53M in Q3, which included 42% year-over-year whole exome and genome test revenue growth



Expanded adjusted gross margins to 48% in the third quarter of 2023



Narrowed adjusted net loss for the third quarter of 2023 to \$21.1 million, an improvement of 70% year-over-year; expecting turn to profitability in 2025



\$164M in *pro forma cash, cash equivalents, marketable securities and restricted cash* as of 9/30/23; after \$48.8M net financing proceeds received on October 27, 2023

# Test Mix Continues to Shift with Growth in WES/WGS Volume

	3Q23	2Q23	1Q23
<b>Volumes</b>			
Whole Exome, Whole Genome	13,216	11,855	8,705
Exome based Panels	2,922	3,472	3,136
Hereditary Cancer	8,556	7,142	7,120
Other individual gene tests and multi-gene disease panels	32,939	32,459	33,817
<b>Total</b>	<b>57,633</b>	<b>54,928</b>	<b>52,778</b>

**WES/WGS tests grew to 13,216**  
*+71% year-over year and +11% vs Q2*

The unadjusted and adjusted results from continuing operations for the applicable quarters include the combination of results from the Legacy GeneDx diagnostic business with the data and information business of Legacy Sema4, and exclude the results of the discontinued Legacy Sema4 diagnostic testing business. Actual results include GeneDx's continuing operations and the financial impacts of exited Legacy Sema4 business activities.



# Revenue Growth Driven by WES/WGS

	3Q23	2Q23	1Q23
<b>Revenue from continuing operations (\$ millions)</b>			
Whole Exome, Whole Genome	\$ 34.0	\$ 28.7	\$ 22.4
Exome based Panels	1.7	2.0	2.0
Hereditary Cancer	4.5	3.8	4.3
Other individual gene tests and multi-gene disease panels	8.8	8.6	10.6
Data information	1.4	2.1	1.3
<b>Total</b>	<b>\$ 50.4</b>	<b>\$ 45.2</b>	<b>\$ 40.6</b>

**\$34M Q3 WES/WGS revenue**

*+42% year-over year; +62% excluding a one-time benefit in Q3 2022 and +28% QoQ*

*>60% exome adjusted gross margin*

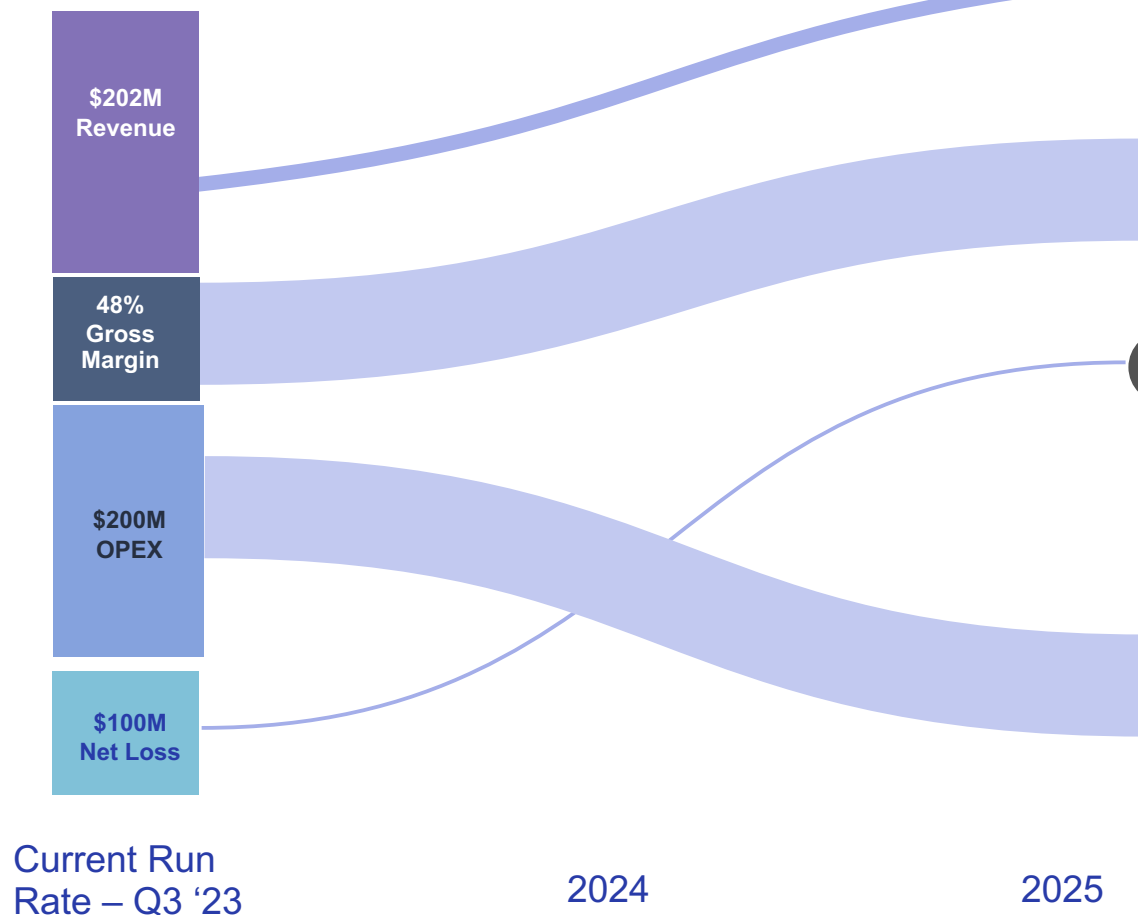
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# Our Path to Profitability

For illustrative purposes, not formal guidance

## Three drivers to sustainable profit



### Revenue Growth

- Revenue growth driven by whole exome/genome testing
- Exome revenues, adjusted for a one-time benefit in Q3 2022, grew 62% year-over-year during Q3 2023
- Revenue breakeven level by end of 2025 is achievable with growth rates well below recent exome volume trends

### Break even point

#### Expanding Gross Margins

- Exome gross margin is > 60% today; as exome continues to grow as a percent of overall testing, total margin trends to >60%
- Scaling back lower margin tests, reducing COGS per test, and increasing collection rates each represent upside to current margin levels

#### Rationalizing Operating Expense

- Removing \$40 million in specifically identified operating costs, with all remaining investments focused on exome volume and revenue growth



# 2023 Guidance

## Updated October 30, 2023

Revenues between \$187-192 million for full year 2023

Expanded adjusted gross margin profile 2023 and beyond

Expects to use \$75 to \$79 million of net cash for the second half of 2023, inclusive of servicing obligations of the previously exited business activities

Expected to turn profitable in 2025



# Appendix

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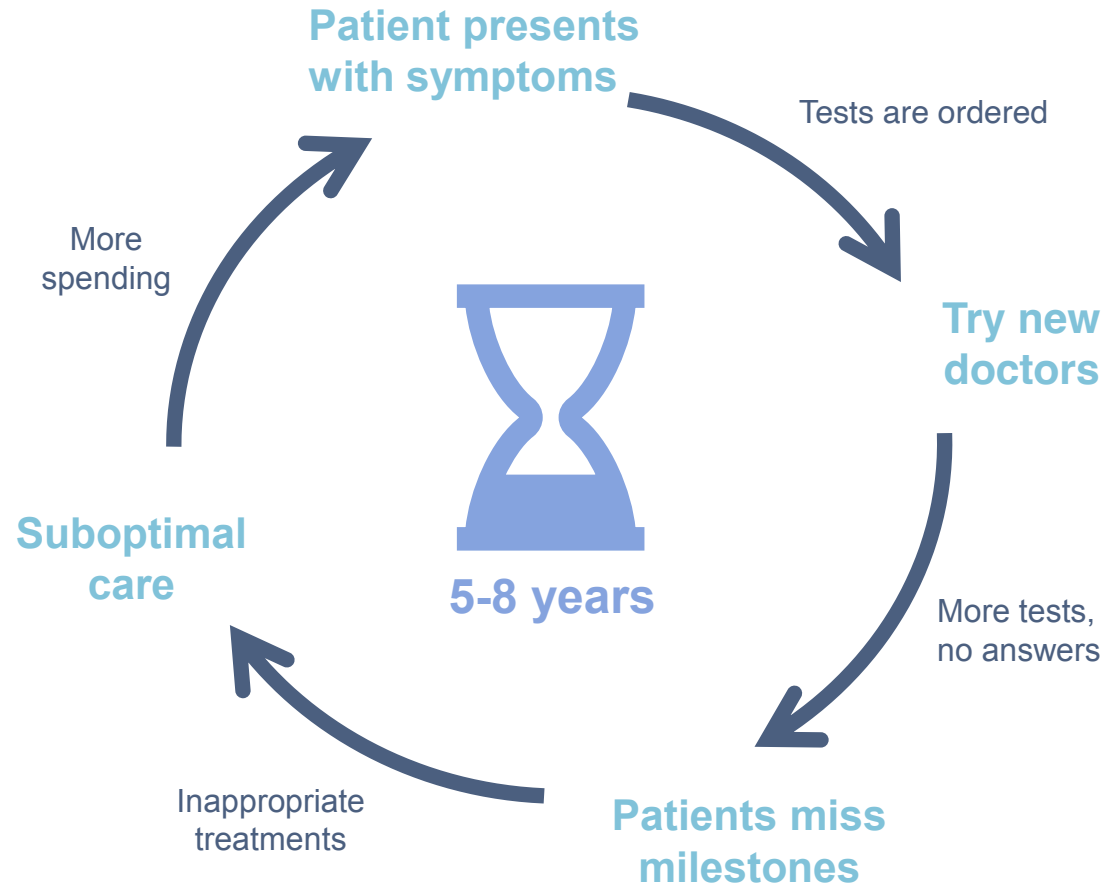


**Our mission:** To deliver personalized and actionable health insights to inform diagnosis, direct treatment, and improve drug discovery

GeneDx

# The problem: Diagnostic odyssey leads to suboptimal care

Lack of genomic data and connectivity can cause a cycle of misdiagnosis



## Missed diagnoses impact everyone

### Patients/Caregivers

- Inappropriate treatments and delayed care
- Suffering

### Healthcare system

- Increased costs and burden
- Missed opportunities to develop treatments

### Providers

- Stress from continued patient suffering
- Inconclusive diagnoses

# Rare disease is a misnomer: collectively, common

A constellation of a multitude of disorders

## Oncology

**>100** types of disease

**1 in 8** people will be diagnosed with breast cancer during their lifetime

**1,300+ therapies**

in clinical testing for cancer

## Rare Diseases

**>7000** individual diseases

**1 in 10 people** affected in the US

**700 medicines**

in development with regulatory pathway via Orphan Drug Act





**Our Solution:**  
**Genomic Data + Clinical Insights**  
**= Better Health**

GeneDx

# GeneDx: Leaders in genomic interpretation and health insights



**Best-in-class genomic expertise**

**20+ years of genomic analysis and pioneering work in identification of new disease-causing genes**

- › **Higher diagnostic yields** compared to multi-gene panels, leveraging internal database and interpretation expertise
- › **Fewer uncertain findings** in disease diagnosis
- › **More than 500,000 clinical exomes and genomes sequenced**, driving an unparalleled genomic database enriched for rare disease



**Centrellis<sup>®</sup> health intelligence platform**

**Proprietary intelligence platform built to ingest and synthesize clinical and genomic data**

- › **Integrates digital tools with artificial intelligence** to deliver better health insights
- › **Ability to combine data** from an electronic medical record and integrate it with genomic data
- › **Delivers context** and a more complete understanding of complex disease

**Our platform provides actionable responses to diseases throughout the course of a patient's life**





# Genomics-based healthcare: the future of medicine

Supported by scientific evidence, literature, clinical practice, and industry guidelines

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 cardio and neuro to replace multi-gene panel and individual gene tests

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

\*Company Estimates supported by DefinitiveHC diagnosis data.

# Enhanced focus on exome and genome sequencing is working

High volume and revenue growth, expanding gross margins, and reduced cash burn

**2022 Financial Results\*** | Excluding revenues and costs from exited business activities, GeneDx has:

**\$171M**

Revenue generated

*A 38% increase from 2021*

**>180,000**

Test results volume

*A 23% increase from 2021*

**39%**

Adjusted gross margin<sup>†</sup>

\*The pro forma unadjusted and adjusted results from continuing operations for 2022 and the comparable results for 2021 are presented on a pro forma basis assuming Legacy GeneDx and the Company were combined for the entirety of 2021 and 2022 and exclude the revenues and costs from the now discontinued Legacy Sema4 diagnostic testing business, and include the combination of the Legacy GeneDx diagnostic business revenues and costs with the data and information revenues and associated costs derived from the Legacy Sema4 business. Actual total Company results include the results of the Legacy GeneDx business only from the date of the Company's acquisition of Legacy GeneDx on April 29, 2022 and the purchase accounting associated with the acquisition of Legacy GeneDx and also include the financial impacts of exited Legacy Sema4 business activities for the full year.

<sup>†</sup>Adjusted gross margin is a non-GAAP financial measure. GeneDx has not provided a reconciliation of its preliminary, unaudited Adjusted Gross Margin to the most directly comparable GAAP measure because certain items excluded from GAAP cannot be reasonably calculated or predicted at this time. Accordingly, a reconciliation is not available without unreasonable effort.

**GeneDx**

# Enhanced focus on exome and genome sequencing is working

High volume and revenue growth, expanding gross margins, and reduced cash burn

**Q2 2023 Financial Results\*** | Excluding revenues and costs from exited business activities, GeneDx has:

**\$45M**

Pro forma revenue generated

*13% increase from Q2 2022*

**~55,000**

Test results volume

*22% increase from Q2 2022*

**37%**

Adjusted gross margin†

\*Pro forma results from continuing operations for GeneDx reported today include the combination of Legacy GeneDx and only the data and information business of Legacy Sema4, and assume Legacy GeneDx was owned for the entirety of 2022. Continuing operations exclude revenues and costs from the now discontinued Legacy Sema4 diagnostics testing business.

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**Driving growth and expanding markets**

GeneDx

# Changing the perception of exome and genome sequencing

GeneDx has spent over a decade solving for limitations of the past

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”



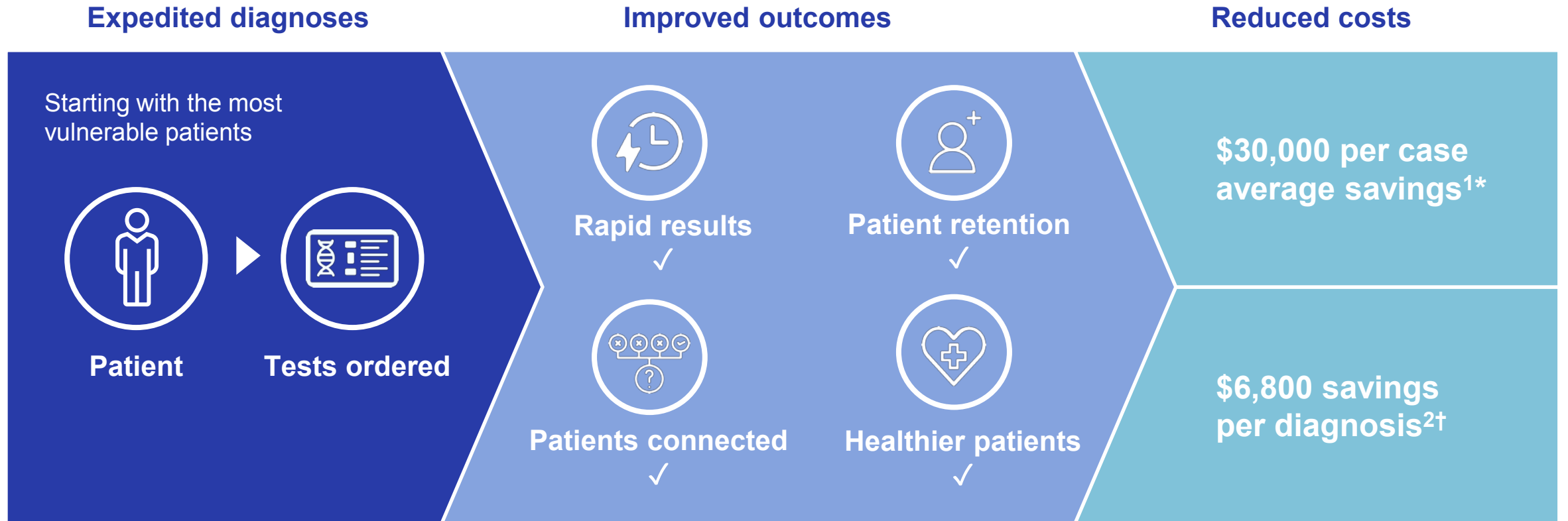
Interpretation

“Results are confusing and filled with useless information”

“My patient can get a definitive diagnosis”

GeneDx

# The inherent value of exome sequencing diagnostics



\*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

# 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



# An ever-growing number of commercial payers and Medicaid programs are adopting favorable coverage<sup>1</sup>

## Commercial coverage for exome and genome

- › **70%** offer coverage when criteria are met
- › **UnitedHealthcare<sup>®</sup>** and **Cigna<sup>®</sup>** cover exome/genome under their commercial plans

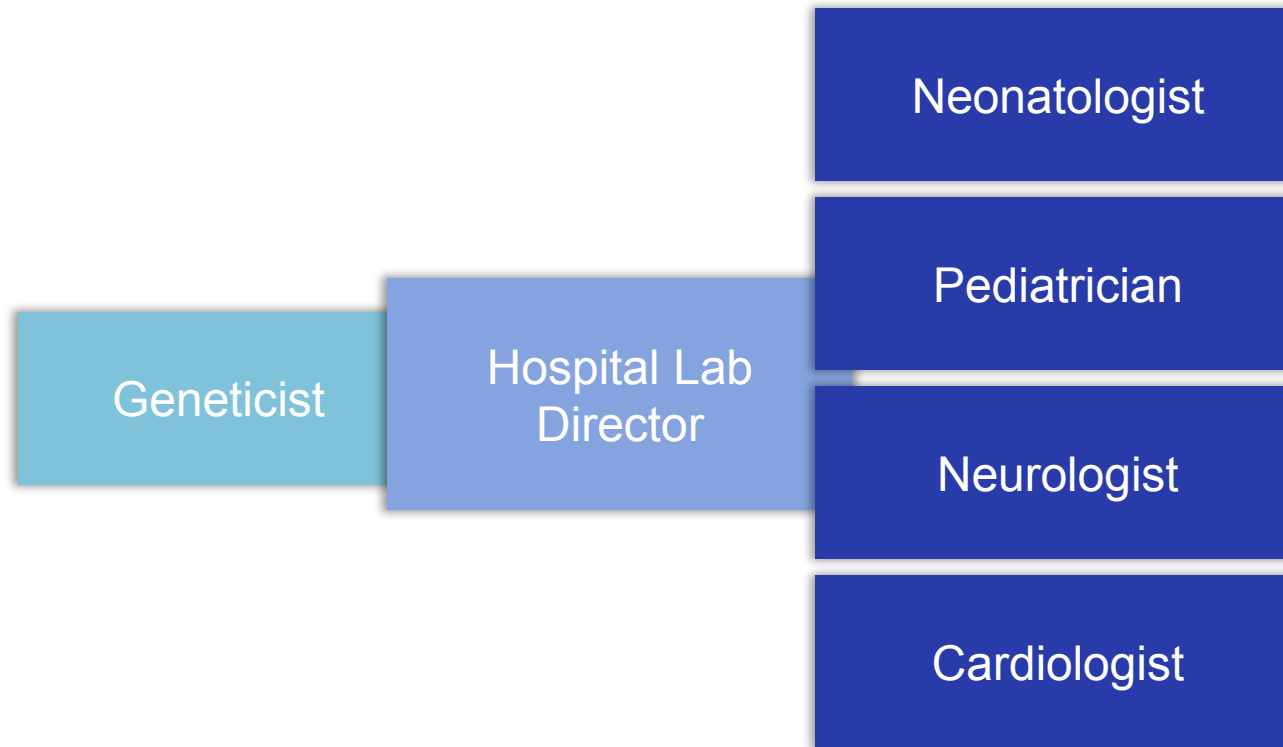


## Medicaid coverage of genomic sequencing

- › **28 states** cover exome sequencing
- › **8 states** cover rapid genome sequencing

Total commercial payer targets represent >93% of commercially insured lives in US. Based on covered life data from DRG database accessed on September 13, 2022.

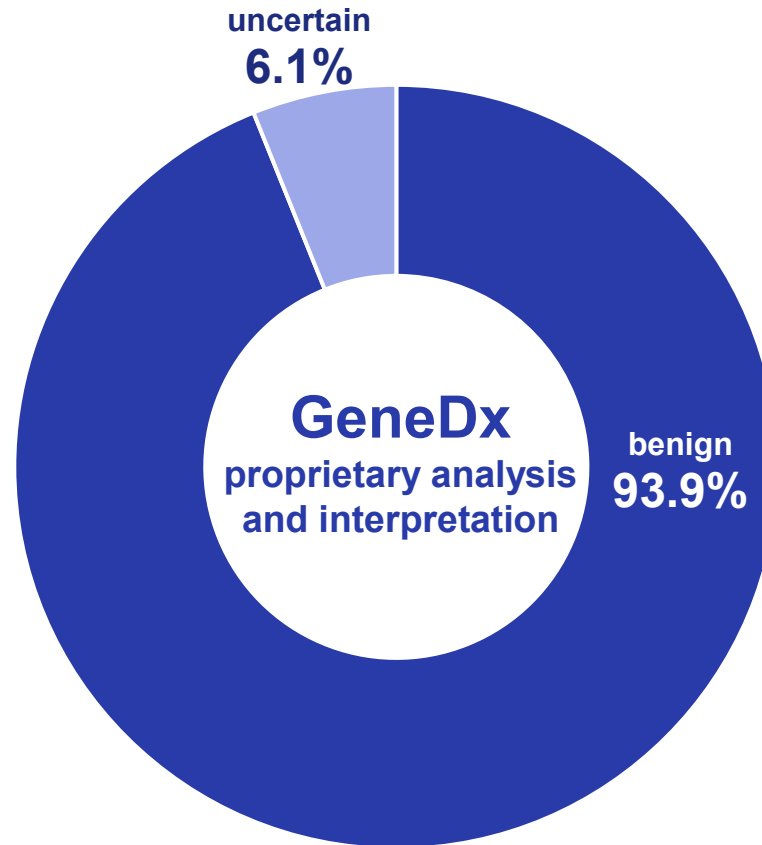
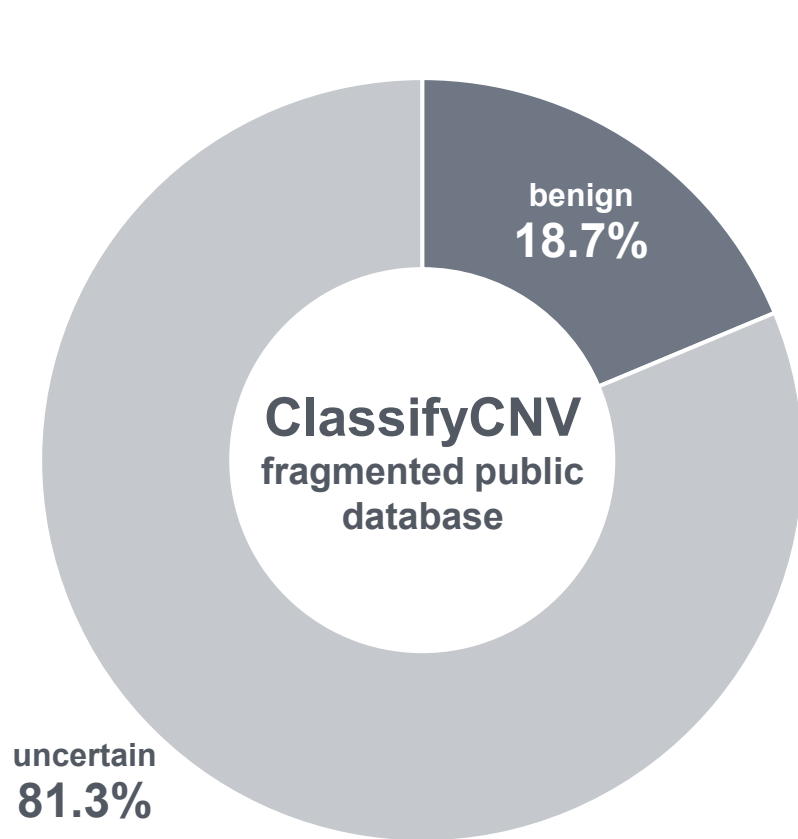
# GeneDx leverages decades of earned trust with geneticists to develop relationships with physicians



We currently have relationships with >500 major health systems

\*LIMS, final reported test volume and # of new ordering clinicians, date through June .

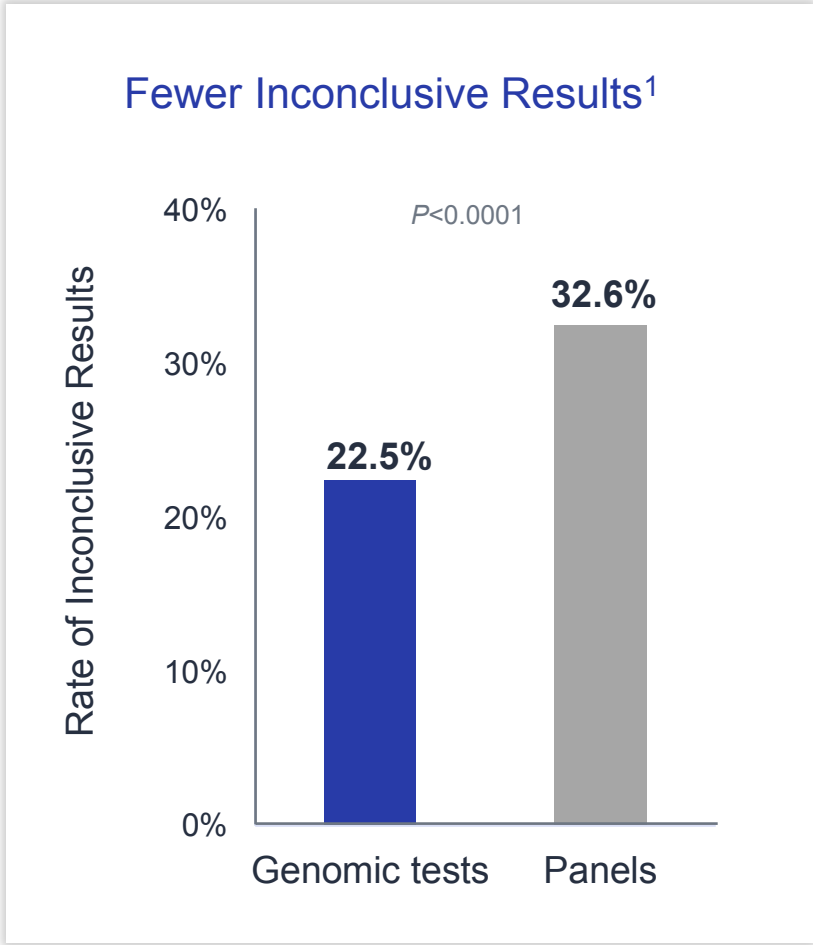
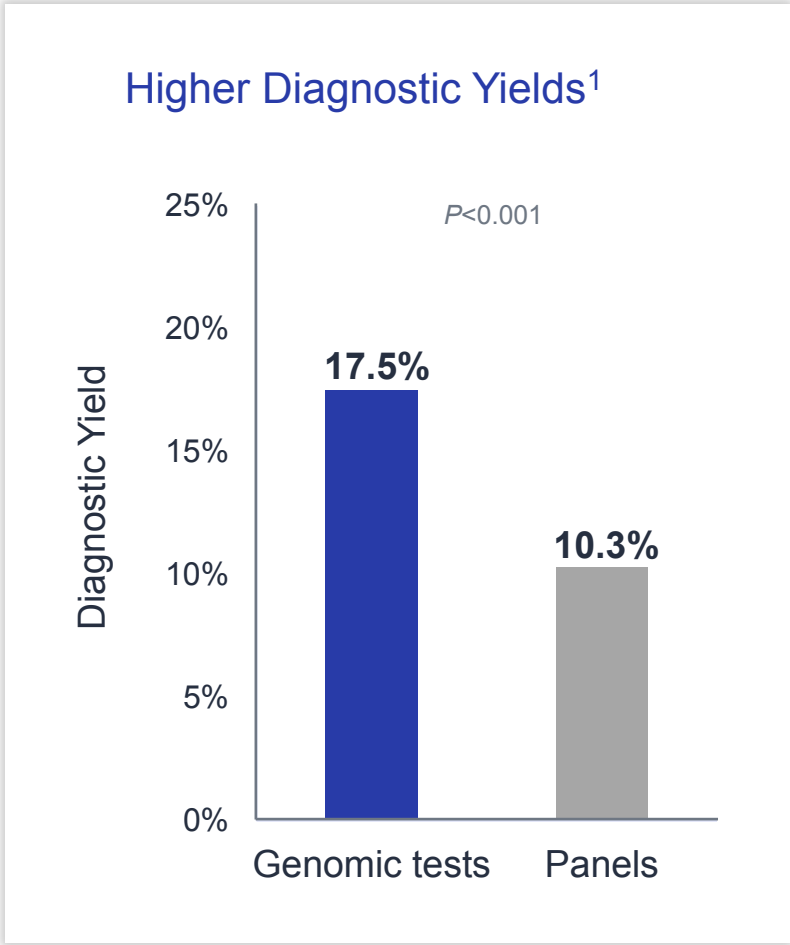
# Unparalleled GeneDx interpretation platform powered by 500,000 exomes delivers fewer uncertain findings



- Fewer uncertain findings lead to improved clarity for clinicians and patients
- More expertly curated disease-causing variants vs public data sets
- Definitive diagnosis in more cases means fewer reviews may be needed

**5x fewer** complex variants for review with GeneDx

# Delivering greater certainty is critical for expanding utilization



- Use of genomic sequencing tests reduced the inconclusive rate and improved diagnostic yield
- Rate of inconclusive results increases with larger multi-gene panel size
- Use of trio sequencing leads to higher diagnostic yield and fewer inconclusive results

Reference: 1. Rehm HL, Alaimo JT, Aradhya S, et al. *MedRxiv*. 2022. <https://doi.org/10.1101/2022.09.21.22279949>

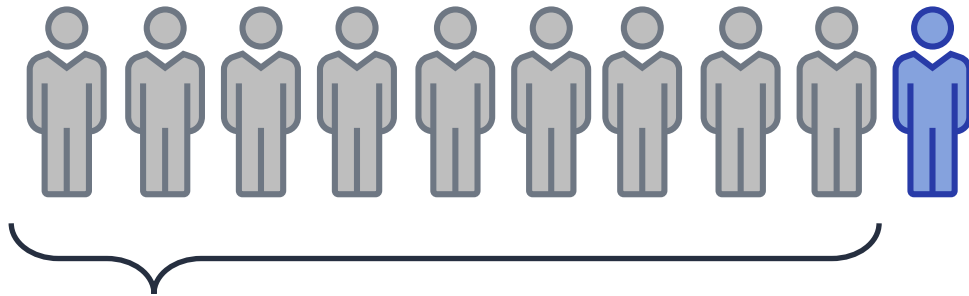


# Panels leave patients behind. Exome and genome do not.

Exome analysis provides higher diagnostic rates and shorter diagnostic wait times than traditional testing strategies

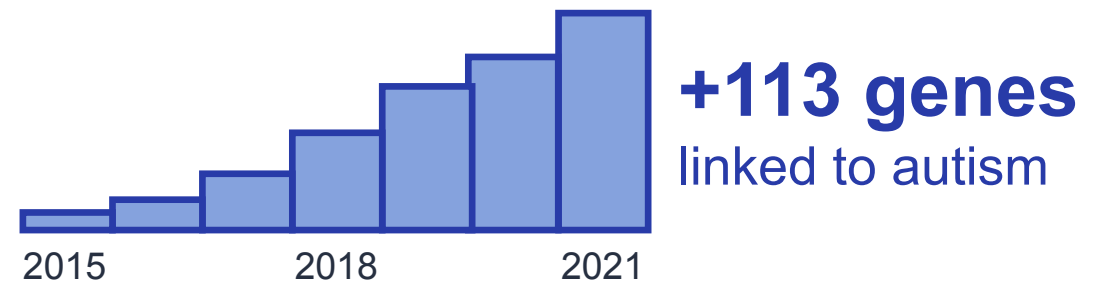
An evaluation of almost 19,000 individuals with autism spectrum disorder revealed:

## Shortcomings of prior genetic testing



**~90%** of the patients had a **previous genetic test with negative results**

## Advances in technology are improving diagnoses



From 2015 to 2021, **113 genes** that had no established connection to autism have been **upgraded to “disease-causing”**

The snowball effect of accumulated **GeneDx** data identifies more pathogenic findings that others miss

**Reference:** Lindy A, Torene R, Retterer K, Kruszka P. Evaluation of 18,911 individuals with autism reveals that exome analysis provides higher diagnostic rates and reduced time to diagnosis than traditional testing strategies. Accepted abstract. American Academy of Neurology Annual Meeting. 2022. Seattle, WA

**GeneDx**

# Exploring innovative methods to enhance diagnostic capabilities and open a new chapter in precision care

Established research collaboration to study the capabilities of HiFi long-read whole genome sequencing to increase diagnostic rates in pediatric patients

**PacBio**

**W**

UNIVERSITY *of* WASHINGTON

Study is first of its kind to compare diagnostic rates across short- and long-read sequencing platforms

**GeneDx**

# We transform the standard of care to meet the standards of the evolving world



SeqFirst is studying the use of rapid whole genome sequencing to improve care and outcomes of children in the intensive care units at Seattle Children's Hospital.



BROTMAN BATY  
INSTITUTE



Seattle Children's  
HOSPITAL · RESEARCH · FOUNDATION



The GUARDIAN study is using genome sequencing to screen for more conditions than those currently included in standard newborn screening.



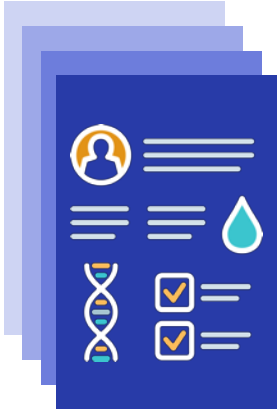




**Building the future of healthcare**

GeneDx

# Building the future of healthcare with our comprehensive tools



## Sequence once

Expand the use of and deliver best in class whole exome/genome analysis & interpretation

- > **500K** clinical exomes
- > **2.7 million** phenotypes
- > **1 million** DNA extractions
- > **400 gene discovery** publications

## Combine clinical + genomic data

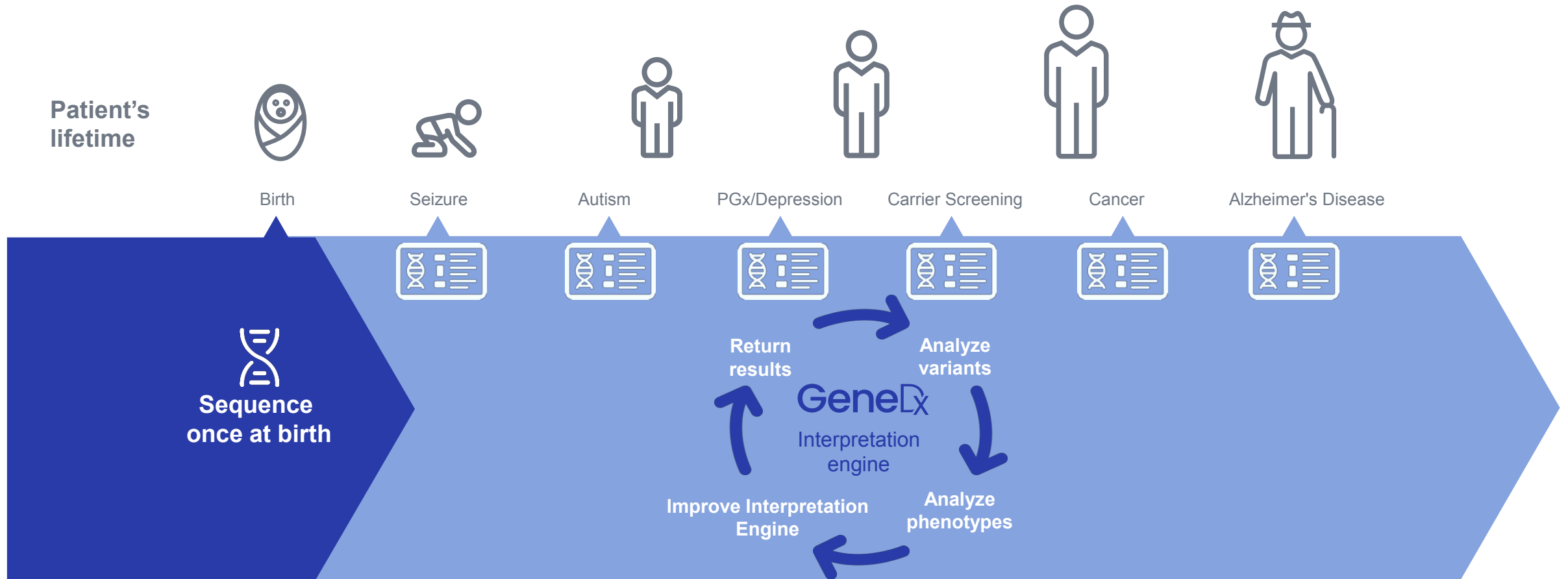
Match structured clinical and genomic data and supplement with disease models to drive insights

- 3.1 million** patient health records
- 8 million** disease diagnoses
- 47 million** phenotypes
- 20 years** of records abstracted from **56 million** clinical documents

## Generation of genomic insights over time

Generate patient-specific and disease profiles to obtain actionable reports upon presentation of symptoms

# Sequence at birth; analyze when needed; deliver actionable information across the lifespan

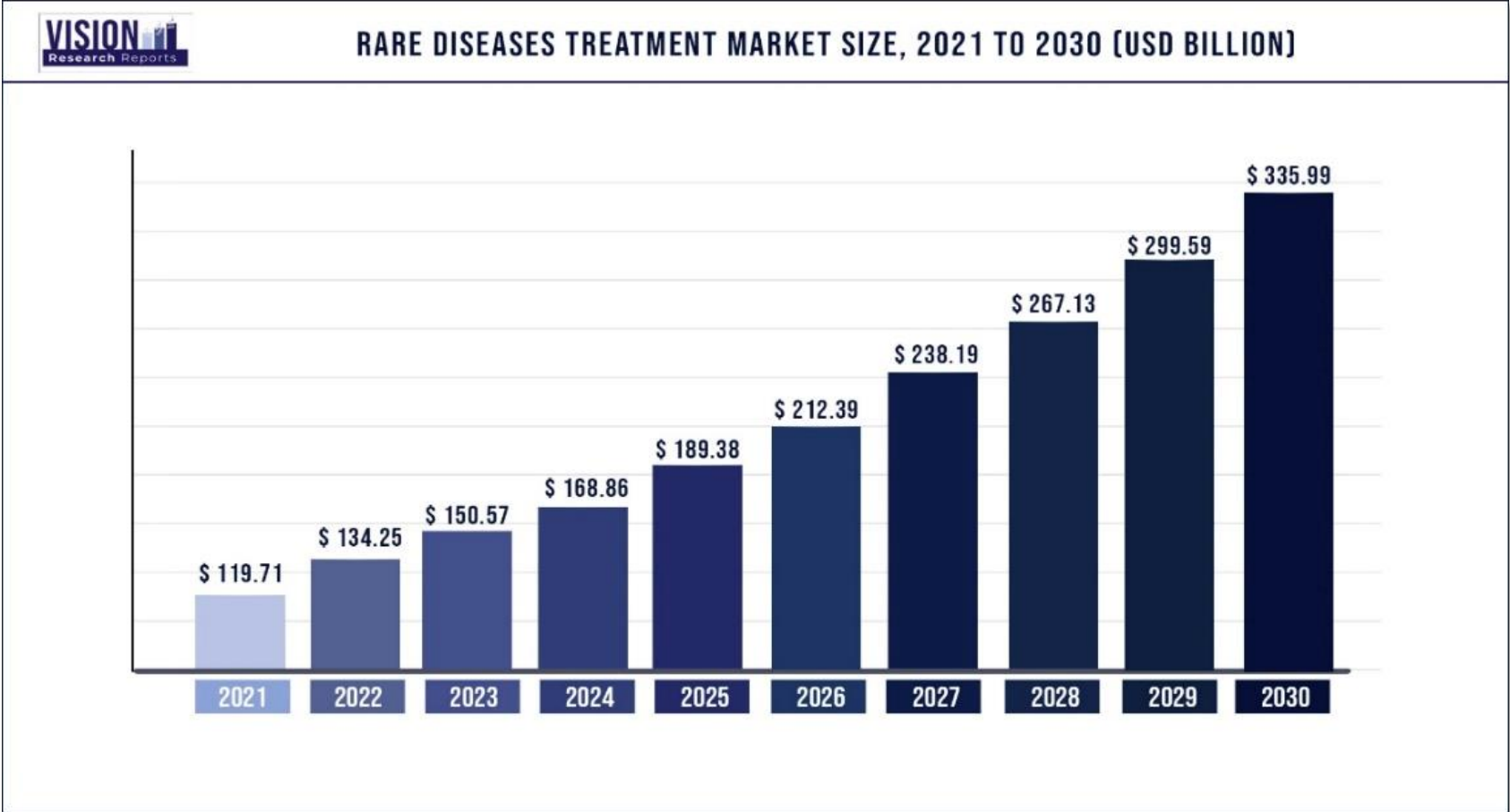


Potential for earlier diagnosis and better outcomes



# Untapped partners: Biopharma

In 2020 alone, more than half (55%) of novel new drug and biological approvals were orphan drugs for rare diseases.



# Data strategy leverages Centrellis as a solutions provider to biopharma partners

Combining clinical and genomic data from patients unlocks upside as platform expands



## DRUG DISCOVERY & EXPANSION

Advance genetically prioritized targets to pre-clinical assets



## PATIENT IDENTIFICATION & OUTREACH

Identify patients appropriate for clinical trials



## REAL WORLD INSIGHTS

Make commercial activities more precise and efficient

## FIND (0-5 year focus)

Repeatable services model for outreach to patients with **Rare Disease** and their clinicians for clinical trial recruitment or delivery of targeted Rx

## UNDERSTAND (1-5 year focus)

Reports and analytics leveraging clinicogenomic data in **Rare Disease** within Centrellis to support R&D for targeted therapies

## PLATFORM (Long term)

SaaS offering: **Therapeutic Area agnostic** access to data, patients, and insights for RWE/RWD to support end-to-end drug discovery pipeline

# Building an ecosystem of partners



**One test. There's no one better.**

GeneDx

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