GeneDx

J.P. Morgan Healthcare Conference
January 2025
San Francisco, California

Forward Looking Statements

This presentation contains forward-looking statements under the meaning of the Private Securities Litigation Reform Act of 1995. Forward-looking statements are statements that do not relate to historical facts and events and such statements and opinions pertaining to the future that, for example, contain wording such as “may,” “might,” “will,” “could,” “would,” “should,” “expect,” “intend,” “plan,” “objective,” “anticipate,” “believe,” “estimate,” “predict,” “potential,” “continue,” “ongoing,” or the negative of these terms, or other comparable terminology intended to identify statements about the future. Forward-looking statements contained in this presentation may include, but are not limited to, statements about: our future performance and our market opportunity, our expectations regarding fourth quarter 2024 and full year 2024 revenue, adjusted gross margin profile and cash burn in 2024. We cannot assure that the forward-looking statements in this presentation will prove to be accurate. Furthermore, if our forward-looking statements prove to be inaccurate, the inaccuracy may be material. These statements involve known and unknown risks, uncertainties and other important factors that may cause our actual results, levels of activity, performance or achievements to be materially different from the information expressed or implied by these forward-looking statements.

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, (iv) our ability to pursue our new strategic direction, and (v) our ability to enhance our artificial intelligence tools that we use in our clinical interpretation platform. The information, opinions and forward-looking statements contained in this announcement speak only as of its date and are subject to change without notice.

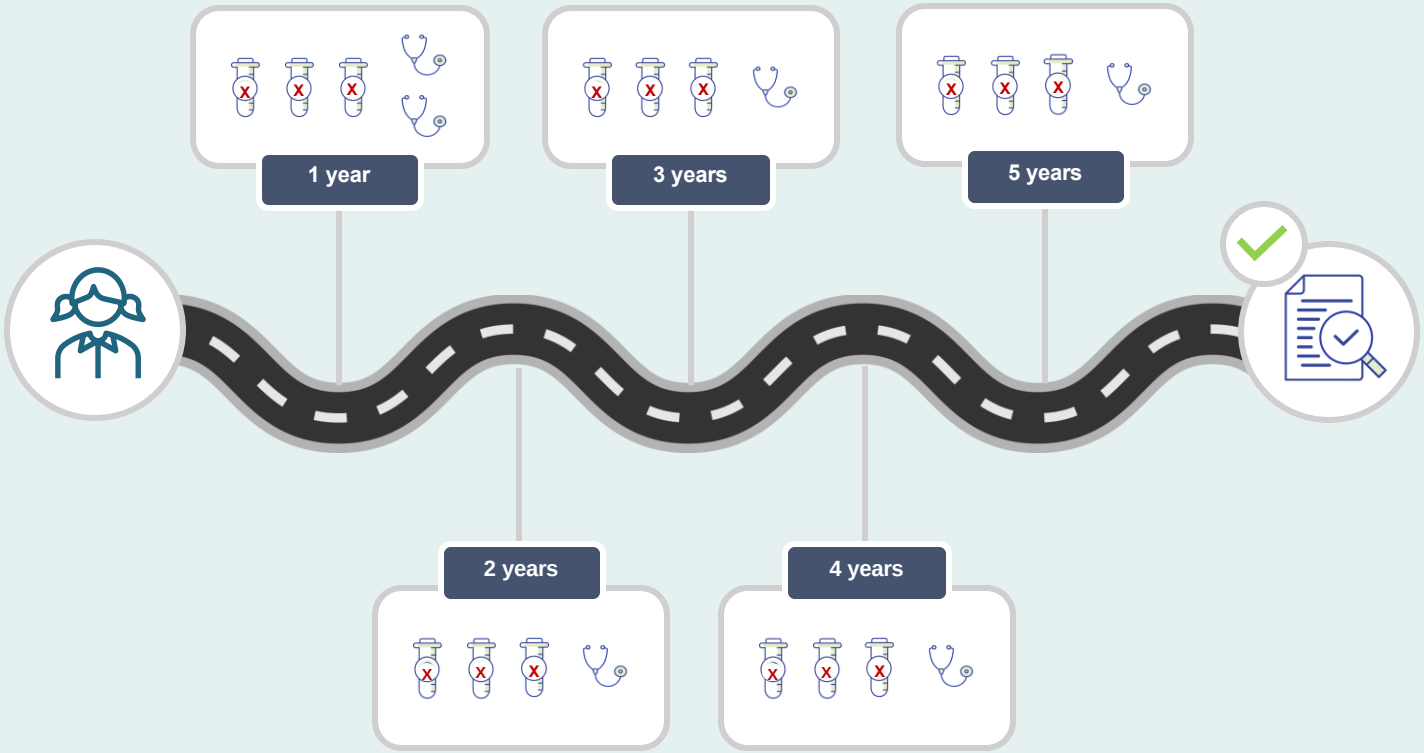
This presentation contains estimates, projections and other information concerning our industry, our business, and the markets for our products and services. Information that is based on estimates, forecasts, projections, market research or similar methodologies is inherently subject to uncertainties, 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. Requests for copies of such documents should be directed to our Investor Relations department at GeneDx Holdings Corp. 333 Ludlow Street, North Tower 6th Floor, Stamford, Connecticut, 06902. Our telephone number is 888-729-1206.

We envision a world where any genetic disorder is diagnosed quickly to prevent disease progression and ensure long and healthy lives for all.

1 in 10 families face an unnecessary diagnostic journey

On average**: 16 tests** and **6 years** before an accurate diagnosis



GeneDx can provide an answer in **days**

References: 1. National Organization for Rare Disorders (NORD). Hope for Millions of Children Living With Rare Diseases. Retrieved from https://rarediseases.org/wp-content/uploads/2024/07/NORD-PRV-One-Pager.pdf 2. Willmen, T., Ronicke, S., Gabriel, H., & Wagner, A. D. (2023). *Rare diseases: why is a rapid referral to an expert center so important?*. BMC Health Services Research, 23(1), 904. Retrieved from https://pmc.ncbi.nlm.nih.gov/articles/PMC10463573/ 3.Marshall, D. A., & Spolador, G. (2021). The complexity of diagnosing rare disease: An organizing framework for outcomes research and health economics based on real-world evidence. Current Opinion in Structural Biology, 68, 1-9. Retrieved from https://www.sciencedirect.com/science/article/pii/S1098360021053831

The diagnostic odyssey: common, critical, and costly

Millions of Americans with a rare disease are urgently searching for answers. Most are children.

Every day without a diagnosis is a missed opportunity for patients—and burden the healthcare system as a whole.

The journey to an accurate diagnosis can take over **six years**.[1]

6 years

3x

On their journey to a diagnosis, rare disease patients will be misdiagnosed an **average of three times.**[2]

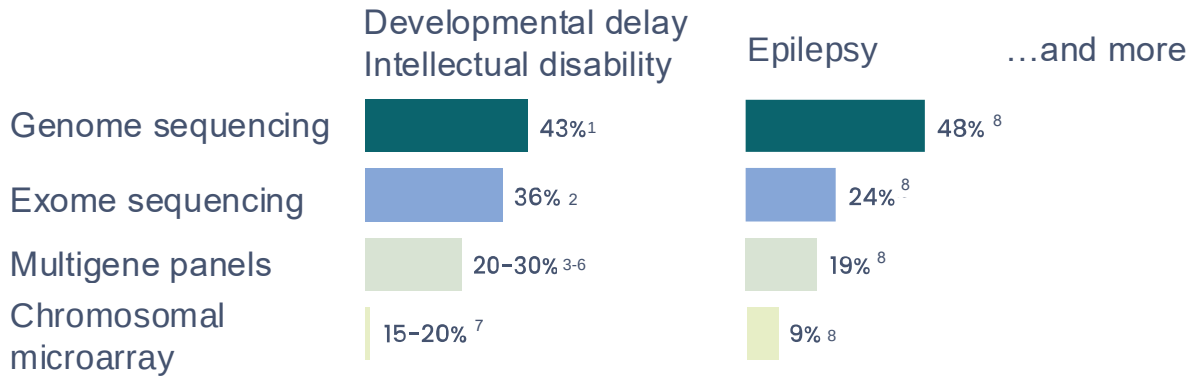
Rare diseases impact **1 in 10 people**, and **over half of them are children.**[3]

The estimated economic burden of rare diseases on the US healthcare system is **nearly $1 trillion** annually.[4]

References: 1. Global Genes. RARE Disease Facts. Retrieved from: www.globalgenes.org/rare-disease-facts/ on June 4, 2024 2. Genetic Alliance UK. The Rare Reality 2016. Retrieved from: https://geneticalliance.org.uk/wp-content/uploads/2024/02/the-rare-reality-an-insight-into-the-patient-and-family-experience-of-rare-disease.pdf. on June 4, 2024 3. National Organization for Rare Disorders (NORD). Hope for Millions of Children Living With Rare Diseases. Retrieved from https://rarediseases.org/wp-content/uploads/2024/07/NORD-PRV-One-Pager.pdf 4. EveryLife Foundation for Rare Diseases. Economic Burden of Rare Diseases in the U.S. Approached $1 Trillion in 2019, Surpassing Cost Estimates for Many Chronic Diseases. Retrieved from https://everylifefoundation.org/economic-burden-of-rare-diseases-in-the-u-s-approached-1-trillion-in-2019-surpassing-cost-estimates-for-many-chronic-diseases/

Exome and genome testing offer answers sooner—leading to more effective treatments and more efficient healthcare spend

Superior diagnostic rate for many indications including:



Recommended by leading medical societies as a first-line test:

American College of Medical Genetics & Genomics (ACMG)
2021 evidence-based guideline[5]

National Society of Genetic Counselors (NSGC)
2022 evidence-based guideline[6]

An earlier genetic diagnosis is proven to:[7,9]

- change medical management
- reduce medical intervention
- result in more timely treatment options
- reduce healthcare costs for patients and the healthcare system
- identify resources and support for parents and family members

References: 1. Manickam K, McClain MR, Demmer LA, et al. Genet Med. 2021 Nov;23(11):2029-2037. doi: 10.1038/s41436-021-01242-6. Epub 2021 Jul 1. **2**. Srivastava S, Love- Nichols JA, Dies KA, et al. Genet Med. 2019 Nov;21(11):2413–2421. doi: 0.1038/s41436-019-0554-6. **3**. Pekeles H, Accogli A, Boudrahem-Addour N, Russell L, Parente F, Srour M. Pediatr Neurol. 2019 Mar;92:32-36. doi: 10.1016/j.pediatrneurol.2018.11.005. **4**. Stefanski A, Calle-López Y, Leu C, et al. Epilepsia. 2021 Jan;62(1):143-151. doi: 10.1111/epi.16755. **5**. Mellone S, Puricelli C, Vurchio D, et al. Front Genet. 2022 Aug 11;13:875182. doi: 10.3389/fgene.2022.875182. **6**. Spataro N, Trujillo-Quintero JP, Manso C, et al. Genes (Basel). 2023 Mar 13;14(3):708. doi: 10.3390/genes14030708. **7**. Savatt JM, Myers SM. Front Pediatr. 2021 Feb 19;9:526779. doi: 10.1186/s13073-022-01026-w. **8**. Sheidley BR, Malinowski J, Bergner AL, et al. Epilepsia. 2022 Feb;63(2):375-387. doi: 10.1111/epi.17141. Epub 2021 Dec 10. **9.** Malinowski, J., Miller, D.T., Demmer, L. et al. Genet Med. 22, 986–1004 (2020). https://doi.org/10.1038/s41436-020-0771-z.

Common diseases are in fact a constellation of genetic diagnoses

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

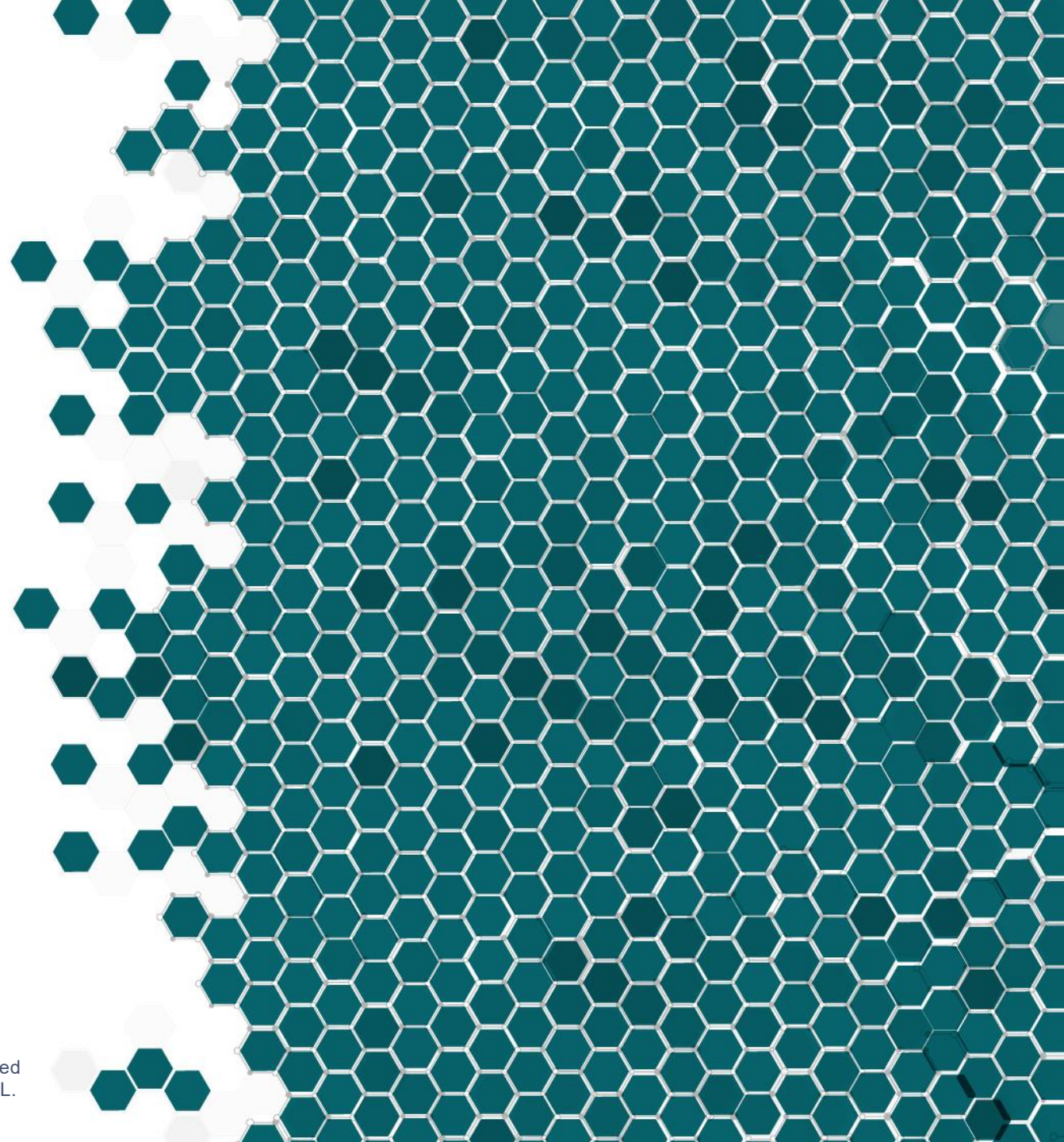
Only 43% of epilepsy genes are tested on many commercial epilepsy panels

Reference: Butler L. *et al.* Exome-based testing for patients with seizures: Advantages over panel-based testing. Poster presented at American Epilepsy Society Annual Meeting; December 2, 2023; Orlando, FL.

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% of epilepsy genes are tested on many commercial epilepsy panels
- Exome and genome sequencing checks all 768 genes



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GeneDx has spent over a decade solving for the limitations of the past and we're working to change the perception of exome and genome sequencing

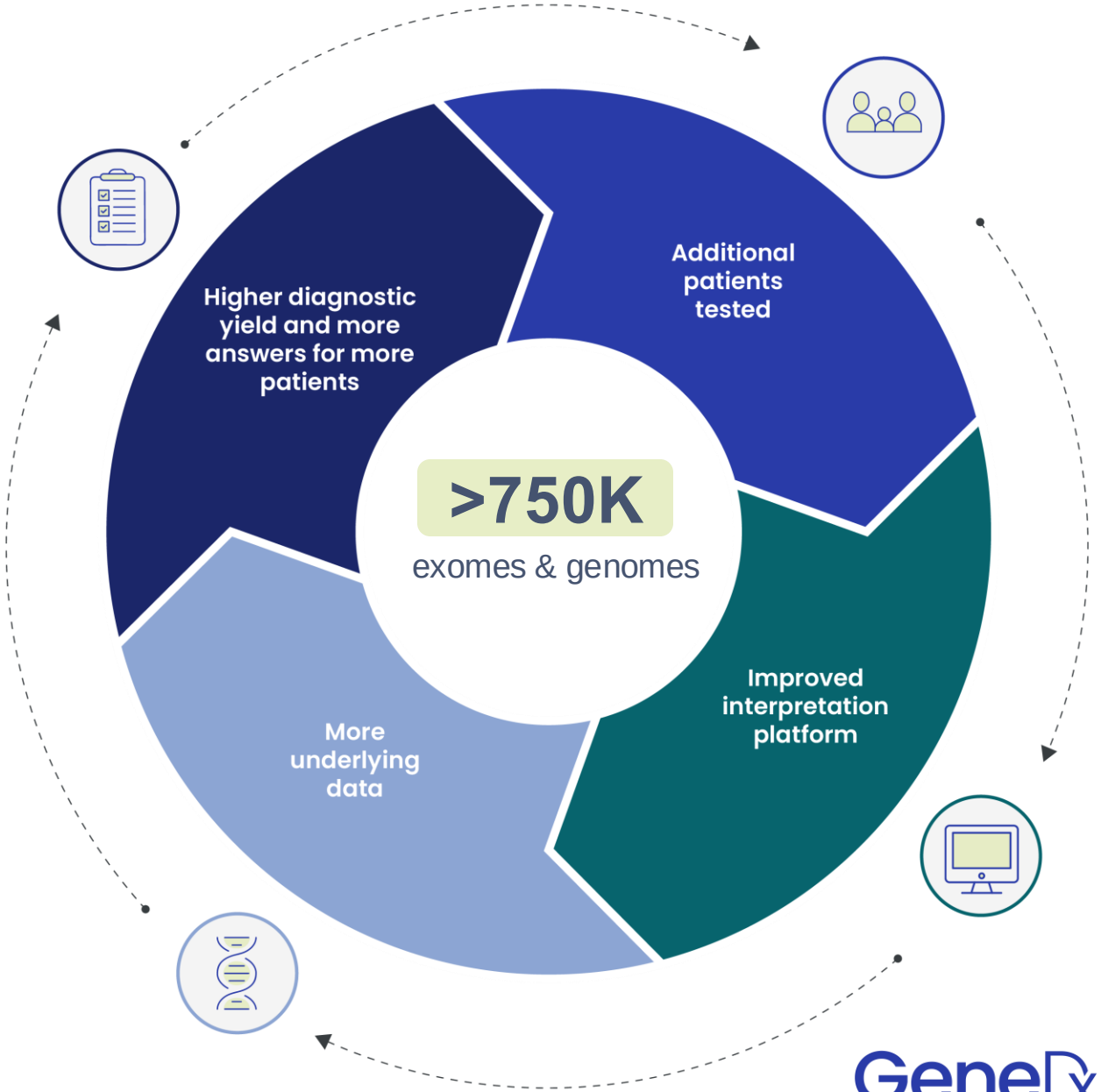
	Then	Now
Turnaround time	Results take months	GeneDx delivers results in days or weeks
Cost	Tests are prohibitively expensive	GeneDx's tests are accessible and widely covered by insurance
Interpretation	Results are confusing, filled with useless information	Patients receive fewer variants of uncertain significance and more definitive answers
Actionability	Nothing to do or change based on the results	Results unlock a growing number of approved therapies, clinical trials, dietary and behavioral health therapies
Value	Other testing (CT scan, MRI, gene panels) offers the same information	Exome and genome uncover what other tests don't, which saves time & money

Accelerating and deepening our competitive advantage with every patient

Pay it forward data strategy: the snowballing effect of data accumulated with ever patient we test drives our underlying interpretation platform to get smarter, faster, and more scalable

That's enable us to identify more than **400 new disease-gene relationships—and counting**.

Patent applications have been filed to develop an IP portfolio directed to our innovative platform of **genetic variant identification, clinical interpretation** and **innovative diagnostic tools** developed using artificial intelligence.

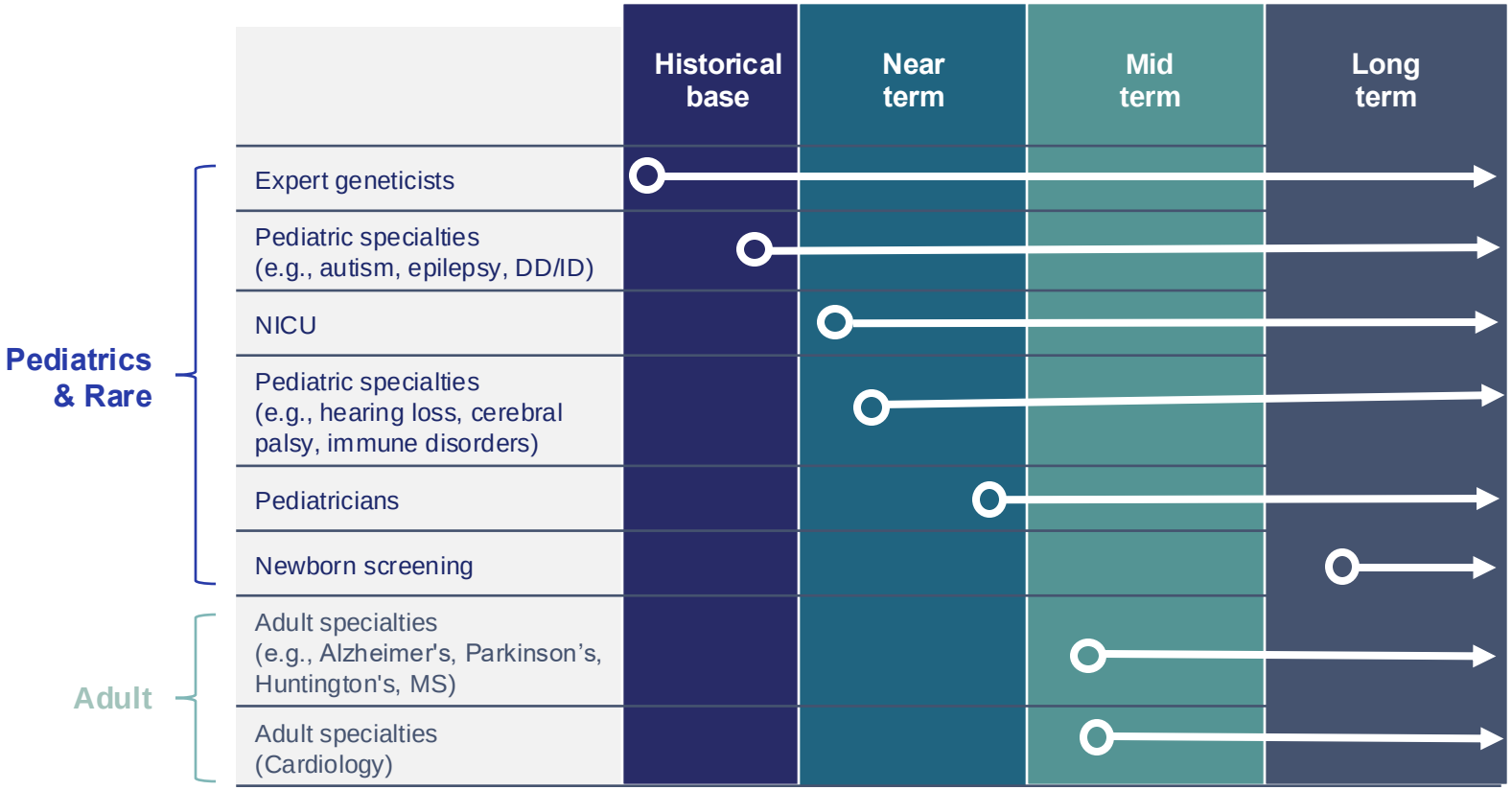


Our data is unmatched in size, breadth, and depth—making it highly infeasible for competitors to recreate

- **Enriched for rare disease**
  Diagnosing even the rarest conditions for 25 years
- **60% of our exomes/genomes are parent/child trios**
  Enabling *de novo* findings, sequencing asymptomatic parents
- **6 million phenotypic datapoints**
  Bridging clinical information and genomic insights
- **10+ years of Medicaid patients tested**
  Representing the full US population diversity
- **All underpinned by expert annotation and curation**
  Bringing answers to more patients today—without future reanalysis

Our market opportunity is massive and poised to expand over time

Taking a disciplined approach entering markets as reimbursement pathways open



Rolling expansion of exome/genome use cases fuels a:

$25 billion ***(pediatrics & rare)***

$20 billion ***(adult)***

$45 billion market opportunity *(total)*

Five-year outlook

Supplemented by a developing biopharma business

Outpatient market expansion: Fueling growth with new indications, coverage and guidelines

Today, GeneDx primarily targets epilepsy, autism and intellectual disability/developmental delay, congenital anomalies, and rare disease

- **We have 80% market share among genetics experts, 12% among pediatric neurologists, and the rest is untapped**

A disciplined approach to expand into **additional indications starting with** hearing loss, cerebral palsy and eventually adult disorders including various neurological, cardiology and other domains

Expect expanded clinical guidelines and reimbursement coverage over time

- **American Academy of Pediatrics (AAP) last updated their genetic testing guidelines in 2014**
- Contracted with ~80% of commercially-insured lives
- Medicaid coverage continues to expand

Inpatient (NICU) market expansion: A clear unmet need, underscored by decades of earned trust and improved workflows

1 in 4 infants in U.S. NICUs likely have a genetic disorder[1]

- Genome testing is severely underutilized, currently **ordered for <5% of children** who could benefit[2]
- NICU orders represent only single digits of our current volume

GeneDx has decades of earned trust amongst children's hospitals and geneticists with 10+ years of exome/genome experience

We are expanding our enterprise sales team and implementing EPIC Aura in 2025 to begin penetrating with a more seamless experience to drive utilization

References: 1. Kingsmore SF, Cakici JA, Clark MM, et al. *Am J Hum Genet*. 2019 Oct 3;105(4):719-733. doi:10.1016/j.ajhg.2019.08.009. **2.** Kingsmore SF, Nofsinger R. & Ellsworth K. *NPJ Genom. Med. 2024*;9(17). doi:10.1038/s41525-024-00404-0.

Genetic evidence is one of the most powerful tools to improve the therapeutic development process

Today

The average therapeutic development cycle:

Costs $2.6B

- Up to 60% of this cost is spent in the clinical phase.

Takes 12 years

- Inefficient trials—driven by patient identification and recruitment challenges—extend timelines.

Fails 90% of the time

- Most trials fail due to safety and efficacy concerns.

Tomorrow

Including genetic evidence can dramatically reduce cost, shorten timelines, and improve success rates:

Genetic evidence can **reduce development costs by up to 25%**.

Leveraging genetic data can **cut development timelines by up to 5 years**.

Drugs with supporting genetic evidence are **2.6 times more likely to succeed**.

Our partnerships with biopharma companies help accelerate treatments—from early discovery through commercialization

Our collaborations are impacting the lives of patients today:

Akouos (Eli Lilly)

GeneDx partnered with Akouos to match patients with the clinical trial that enabled Aissam Dam to hear for the first time.

The New York Times

Gene Therapy Allows an 11-Year-Old Boy to Hear for the First Time

After receiving treatment, Aissam said: "There's no sound I don't like. They're all good."

Regeneron

Through a data partnership with GeneDx, Regeneron received valuable insights into the landscape of hearing loss patients and their associated variants.

The Washington Post
Democracy Dies in Darkness

Deaf baby hears for the first time after 'groundbreaking' gene therapy trial

Opal heard her mother's voice for the first time after participating in Regeneron's clinical trial.

GeneDx

We believe in a future where every newborn's genome is sequenced at birth

Every year, thousands of newborns with actionable conditions are missed by traditional newborn screening (NBS).

Federal NBS guidelines recommend testing for 37 conditions with biomarkers—measurable changes in the baby's blood that indicate the baby may have a disorder.

However, there are hundreds of actionable conditions that lack biomarkers.

Genomic sequencing can detect conditions without biomarkers, expanding the number of conditions screened to ~450.

By supplementing traditional NBS with genomic sequencing, we can offer crucial information to improve health outcomes.

GeneDx is the leader set to revolutionize the standard approach to today's newborn screening, enabling diagnoses before symptoms even start

GeneDx has screened more newborns than any other commercial laboratory. This experience gives GeneDx a deep understanding of how to offer this testing at scale.

Screened 17,000 healthy infants with genome sequencing, toward goal of >100,000

Without this screening, the average age of diagnosis for these conditions is **7-11 years old**

3.2% true positive rate, and **92%** of true positives would not have been detected with today's standard newborn screening

More than **70% of parents consented to gNBS,** with 90% of those opting for inclusion of optional neurodevelopmental disorders

Early diagnosis for conditions like long QT syndrome and Wilson disease **not included in standard newborn** screening, **resulted in life-saving treatments**

Results from the GUARDIAN study, published in ***Journal of the American Medical Association***, set the foundation for clinically-actionable, ethical and responsible gNBS

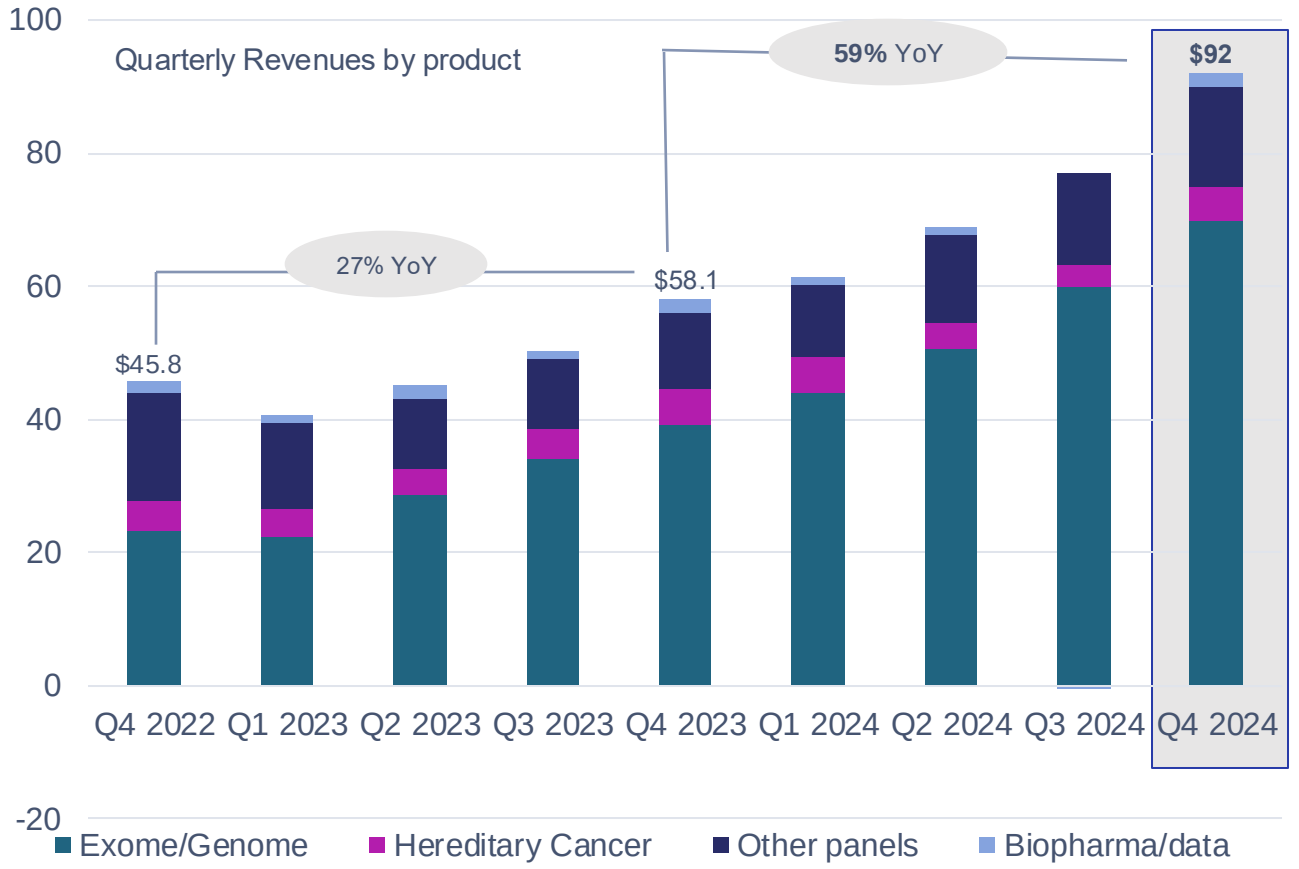
References: 1. Ziegler A, Koval-Burt C, Kay DM, et al. Expanded Newborn Screening Using Genome Sequencing for Early Actionable Conditions. JAMA. Published online October 24, 2024. doi:10.1001/jama.2024.1966

Preliminary results[1] exceeded guidance: FY 2024 revenue[2] now expected to be $299 million (previous guidance was $284-290 million)

Full year 2024 revenues $299 million
- up 54% YoY
- includes a discrete benefit of $6.8 million in fourth quarter in connection with a multi-year appeal recovery from a single payer

Q4 2024 revenues $92 million
- **up 59% YoY**
- up 20% sequentially
- includes that same discrete benefit of $6.8 million

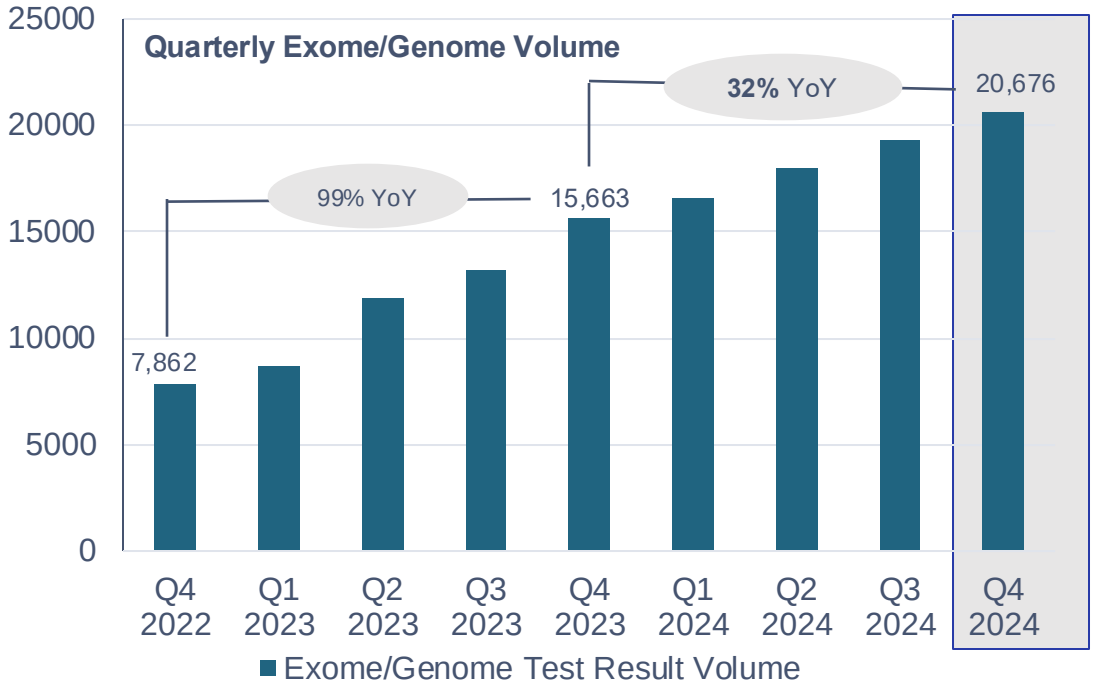
Q4 2024 exome and genome test revenues $75.8 million
- up 93% YoY
- up 26% sequentially
- includes $5.8 million of that discrete benefit

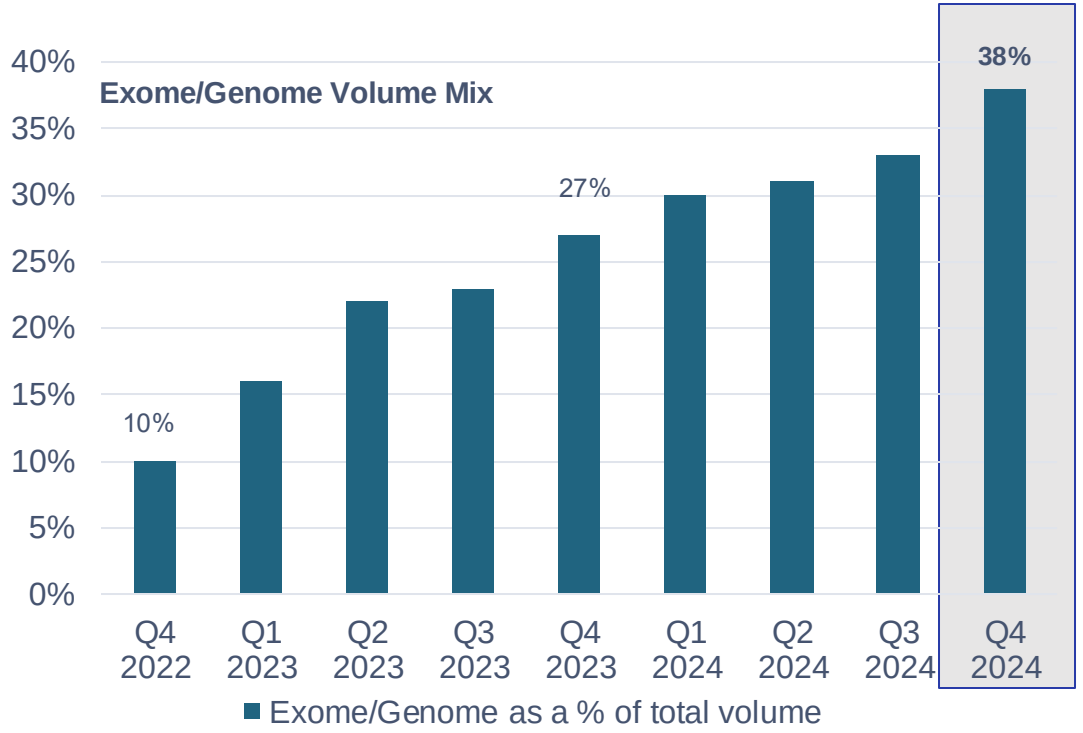


1. GeneDx has not completed the preparation of its consolidated financial statements for the year ended December 31, 2024. The preliminary, unaudited results presented in this presentation for the quarter and year ended December 31, 2024, are based on current expectations and are subject to adjustment, as the company completes the preparation of its 2024 year-end consolidated financial statements and its 2024 year-end audit.
2. Revenue results from continuing operations exclude any revenue from the exited Legacy Sema4 diagnostic testing business.

Preliminary results[1] exceeded guidance: FY 2024 adjusted gross margin[2] now expected to be at least 64% for full year, 68% for Q4 (previous guidance was at least 62% for full year)

- Gross profit benefiting from all three of mix shift towards exome/genome, improved reimbursement and lower production costs
- **Q4 2024 exome/genome volume up 32%** year-over-year and 7% sequentially
- Exome and genome volume mix 38% of all tests' result volume





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2. Adjusted gross margin results from continuing operations, which we believe are representative of our ongoing business strategy exclude any revenue and cost of goods sold from the exited Legacy Sema4 diagnostic testing business. Adjusted gross margin is a non-GAAP financial measures removing depreciation, amortization, share-based compensation and any restructuring costs.

Multiple drivers to profitable, sustainable growth

Expanding serviceable market

- New use cases/ indications / call points stemming from emerging guidelines, expanding and, secular tailwinds towards greater acceptance of exome/genome
- The American Academy of Pediatrics last issued their genetic testing guidelines in 2014. An update in support of an exome/genome first approach for genetics, may unlock the pediatrician call point, of which there are nearly 80 thousand in the U.S.

Driving into the inpatient NICU setting

- SeqFirst and other study data supporting the clinical and economic case for a first-line approach in the NICU
- EPIC Aura go-live Q2 2025

Increasing penetration in outpatient setting

- GeneDx enjoys an ~80% market share of clinical exome/genome ordered in the U.S. today yet we are still only ~12% penetrated in the pediatric neurology market

Reducing denials improving coverage

- Reduction in Medicaid denials via additional states providing exome/genome reimbursement policies
- Reduction in third-party commercial denials through continued refinement of operational processes

New product launches

- Launch additional solutions for biopharma
- Alternative pathways for access and ordering
- Newborn screening (future)

Expanding margins

- Further cost per test declines via introduction of automation/AI across various dry-side processes
- Leverageable commercial spend

Strong capital base

- Turned adj. EBITDA profitable in Q3 2024
- Expecting Q4 2024 to deliver our second consecutive quarter with adjusted net income and our first quarter of positive operational cash flow

A rare opportunity to fuel seismic healthcare shifts

From years of disease progression

From unnecessary and bloated health costs

From generalized treatments

From diagnosing symptomatic disease

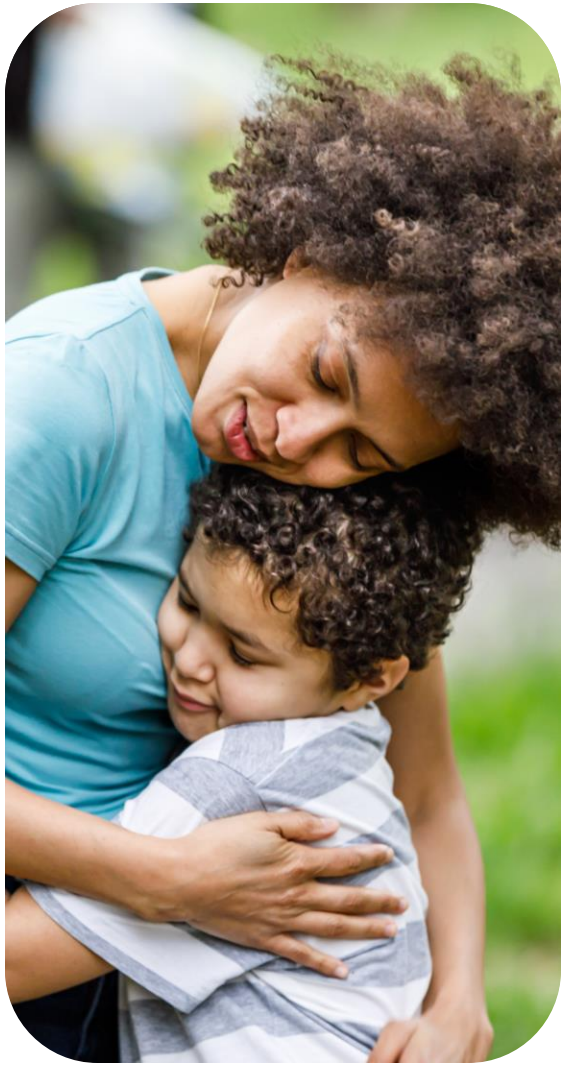
to

early interventions

streamlined economic efficiency

precision medicines

universal genomic newborn screening



We all know the pain of being “too late”

At GeneDx, we’re making sure that children get answers right on time.

We're just getting started.

Thank you

GeneDx