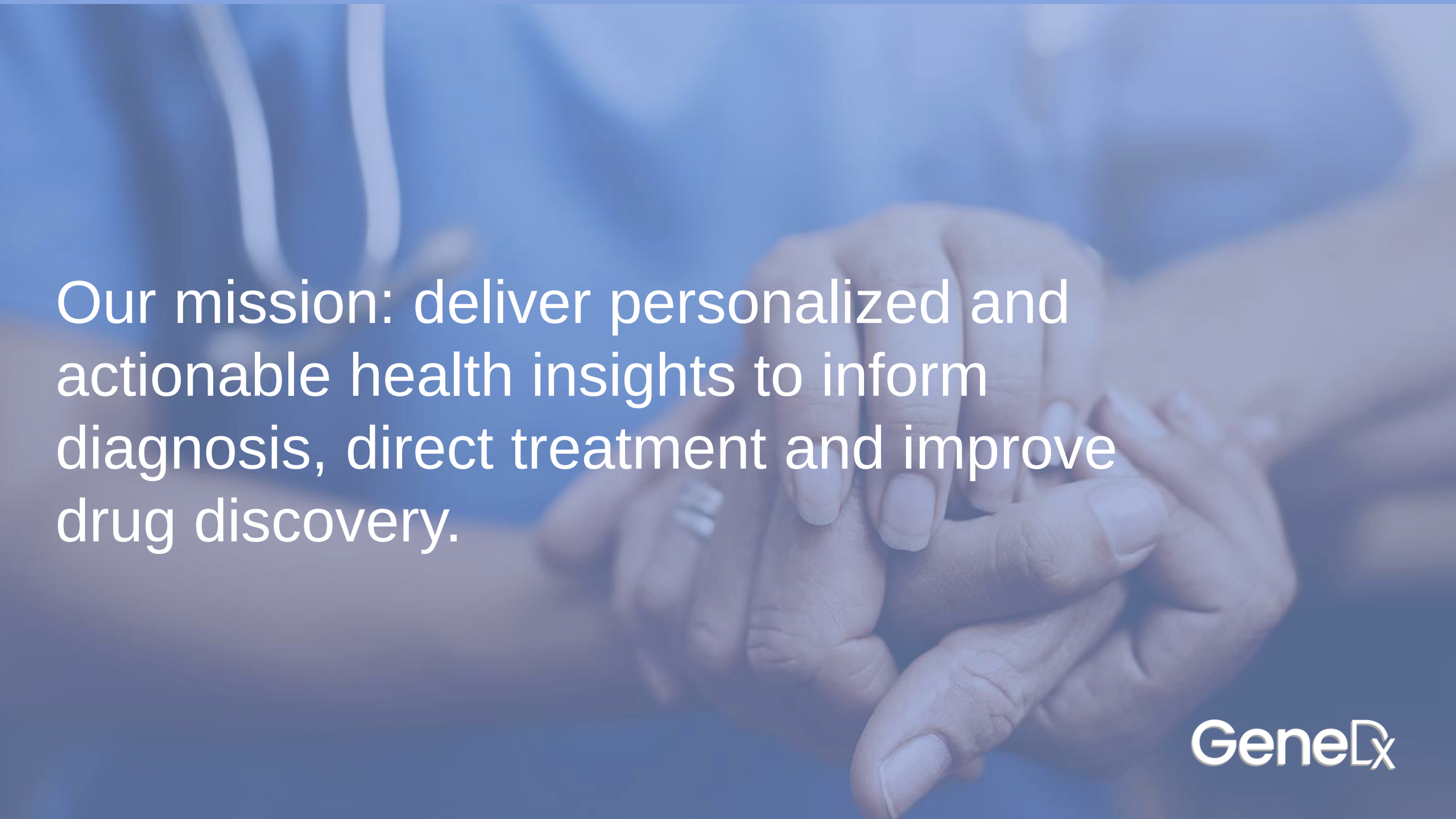
GeneDx: A trusted partner for patient advocacy organizations

Paul Kruszka, MD, MPH, FACMG
Chief Medical Officer
Ultragenyx RARE Bootcamp
November 2024

GeneDx

Who is GeneDx and what do we do?


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

At GeneDx, our goal is to provide clear, accurate, and meaningful answers so that healthcare providers, patients, families, and partners can take action.

National Institutes of Health

Founded in 2000 by geneticists from the National Institutes of Health (NIH) to address the needs of patients diagnosed with rare disorders and the clinicians treating these conditions.

100+ MDs/ PhDs

35 Fellows of ACMG

150+ genetic counselors

1000+ peer-reviewed publications

A global leader in genomic analysis

>650K clinical exomes and genomes sequenced since 2012[1]

One of the largest rare disease datasets

>80% exome market share among ordering clinicians the US[2]

GeneDx

1. As of July 30, 2024 GeneDx laboratory operations attests to having sequenced more than 650,000 clinical genomes and exomes.
2. Based on Definitive Healthcare commercial claims data (2023 through July) for CPT codes 81415, 81416 for clinicians specializing in pediatric medicine & developmental pediatrics

Why is exome testing important? Does insurance pay for it?

GeneDx

An exome-first approach is recommended by experts

For patients with developmental delay, intellectual disability, congenital anomalies, epilepsy, cerebral palsy, or autism spectrum disorder, exome should be the first test offered.[1-4]

American College of Medical Genetics and Genomics (ACMG) recommends exome as a first-line test for developmental delay, intellectual disability, and congenital anomalies (July 2021)[2]

National Society of Genetic Counselors (NSGC) recommends exome as a first-line test 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]

1. Srivastava S, Love-Nichols JA, Dies KA, et al. Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genet Med. 2019 Nov;21(11):2413–2421; https://doi.org/10.1038/s41436- 019-0554-6
2. Manickam K, McClain MR, Demmer LA,et. al; ACMG Board of Directors. 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. PMID: 34211152.
3. 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. 2023 Apr;32(2):266-280. doi: 10.1002/jgc4.1646. Epub 2022 Oct 24. PMID: 36281494.
4. Srivastava S, Lewis SA, Cohen JS, et al. Molecular Diagnostic Yield of Exome Sequencing and Chromosomal Microarray in Cerebral Palsy: A Systematic Review and Meta-analysis. JAMA Neurol. 2022 Dec 1;79(12):1287-1295. doi: 10.1001/jamaneurol.2022.3549

Diagnostic rate by testing option

The diagnostic yield of exome and genome testing is greater than CMA[2,3], and rapid testing turns around results in 5-7 days

~10%

Traditional panel diagnostic yield[1]

~10-20%

CMA diagnostic yield[2,3]

~40%

Exome sequencing diagnostic yield[2]

~40-50%

Genome sequencing diagnostic yield[4-6]

1 Rehm H, Alaimo JT, Aradhya S, et al. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. Genet Med. 2023 Jul 30;100947. doi: 10.1016/j.gim.2023.100947.
2 Srivastava S, Love-Nichols JA, Dies KA, et al. Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genet Med. 2019 Nov;21(11):2413–2421; https://doi.org/10.1038/s41436- 019-0554-6
3. Savatt JM, Myers SM. Genetic Testing in Neurodevelopmental Disorders. Front Pediatr. 2021 Feb 19;9:526779. doi: 10.3389/fped.2021.52679. eCollection 2021. doi: 10.3389/fped.2021.526779
4. Lunke S, Bouffler SE, Patel CV et al. Integrated multi-omics for rapid rare disease diagnosis on a national scale. Nat Med. 2023 Jun 8. doi: 10.1038/s41591-023-02401-9.
5. Maron JL, Kingsmore S, Gelb BD, et al. Rapid Whole-Genomic Sequencing and Targeted Neonatal Gene Panel in Infants With a Suspected Genetic Disorder. JAMA. 2023 Jul 11;330(2):161-169. doi: 10.1001/jama.2023.9350.
6.Dimmock D, Caylor S, Waldman B, et al. Project Baby Bear: Rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care. Am J Hum Genet. 2021 Jul 1;108(7):1231-1238. doi:10.1016/j.ajhg.2021.05.008. Epub 2021 Jun 4.

Exome sequencing is covered

Over **70% of commercial health payers** cover exome sequencing when criteria are met [1]

Clinical criteria and individual coverage requirements vary by health insurer, and we encourage all providers to refer to the specific insurance plan medical policy to review the requirements.

Patients must meet the condition-specific clinical criteria for ES coverage stated in their insurance medical policy.

For some patients, ES may have better commercial insurance coverage than multi-gene panels.

1 Internal review of ~100 commercial payer targets who represent >93% of commercially insured lives in US. Based on covered life data from DRG database accessed on September 13, 2022.

Disclaimers:

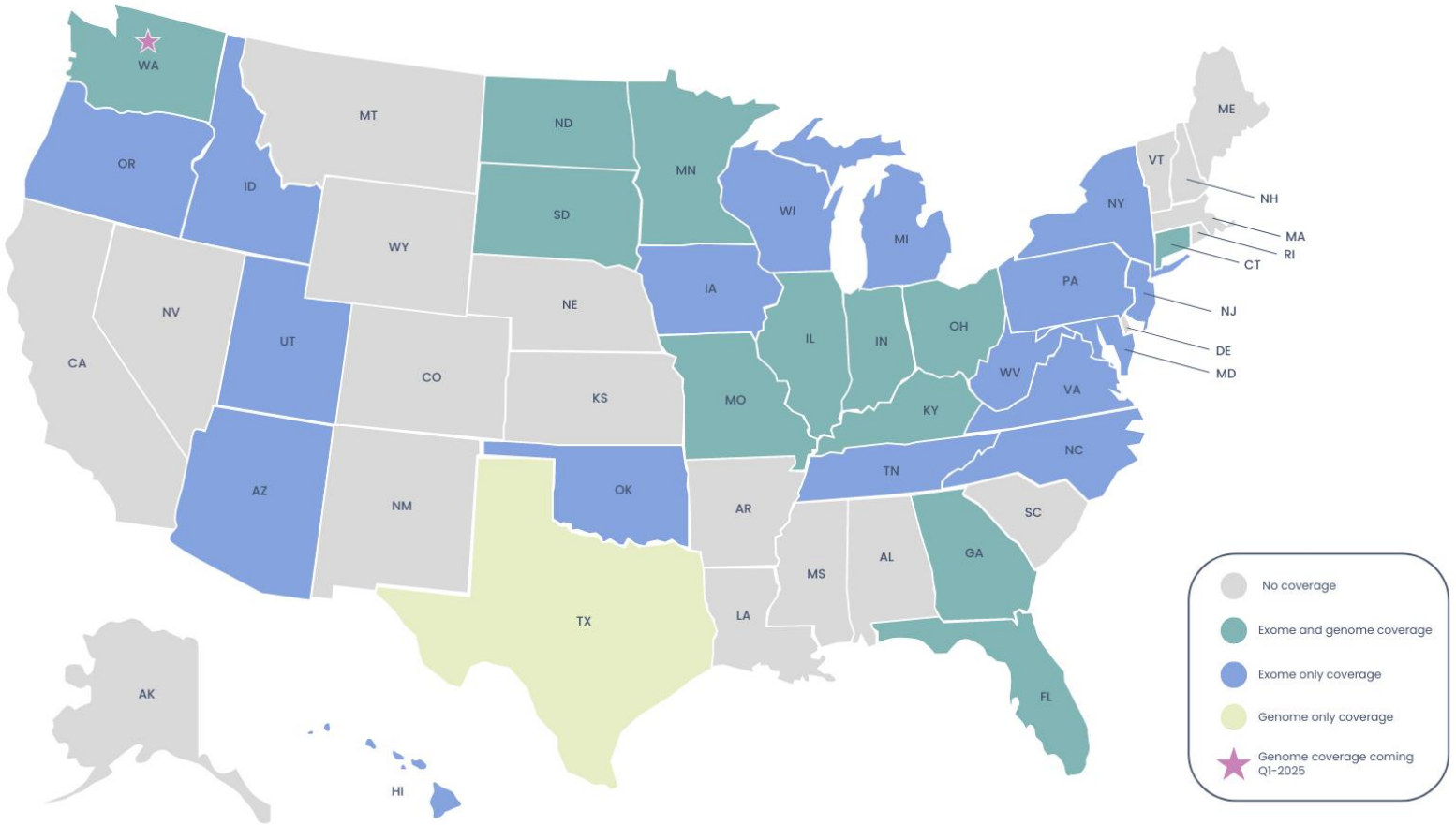
Patient coverage for services and out-of-pockets costs are subject to health plan benefits, medical policy, and any exclusions that may apply. Ordering providers and patients are encouraged to check the patient's health plan directory or inquire with their health plan for network status of any provider for which they are ordering services, ahead of those services being rendered. Health plans may maintain separate medical policies for different insurance products.

All information provided is as of February 1, 2023.

GeneDx

Medicaid coverage for exome and/or genome sequencing in the outpatient setting

30 states have Medicaid coverage for exome and/or genome sequencing in the outpatient setting



Updated 10/04/2024, SS

GeneDx connects newly diagnosed patients & their families to resources.

This page is included with every test report and encourages families to find support & resources.

Resources

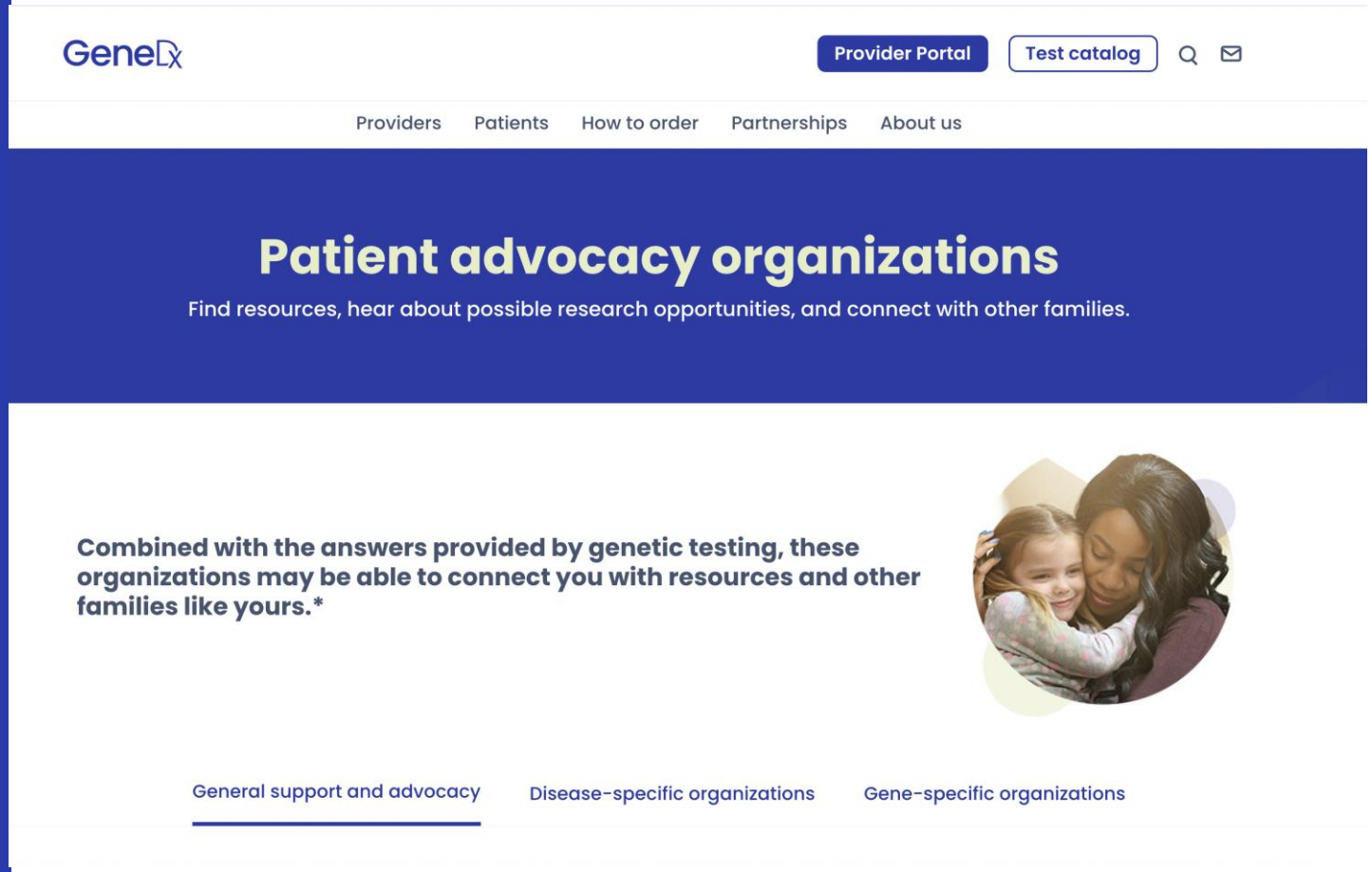
Combined with the information provided by your genetic test report, these groups can connect you with resources and other families like yours. *

For condition- and gene-specific advocacy groups, please visit GeneDx.com/advocacy

- **General support and advocacy**
  - Child Neurology Foundation
    www.ChildNeurologyFoundation.org
    (859) 551-4977
  - Global Genes
    www.GlobalGenes.org
    (949) 248-7273
  - National Organization for Rare Disorders
    www.RareDiseases.org
    (617) 249-7300
- **Information exchange and connection**
  - GenomeConnect: www.GenomeConnect.org
  - MyGene2: www.MyGene2.org

*These clinician and family resources are of an informational nature only. While GeneDx makes every effort to ensure its accuracy, this information is not meant to endorse a particular group, be a complete list, nor serve as medical advice. GeneDx presents this opportunity as a courtesy to clinicians and families who may wish to obtain more details. All GeneDx genetic test reports include this Resource page, no matter the positive or negative result.

The “Resources” page on the report points to this webpage, which has more detailed information.

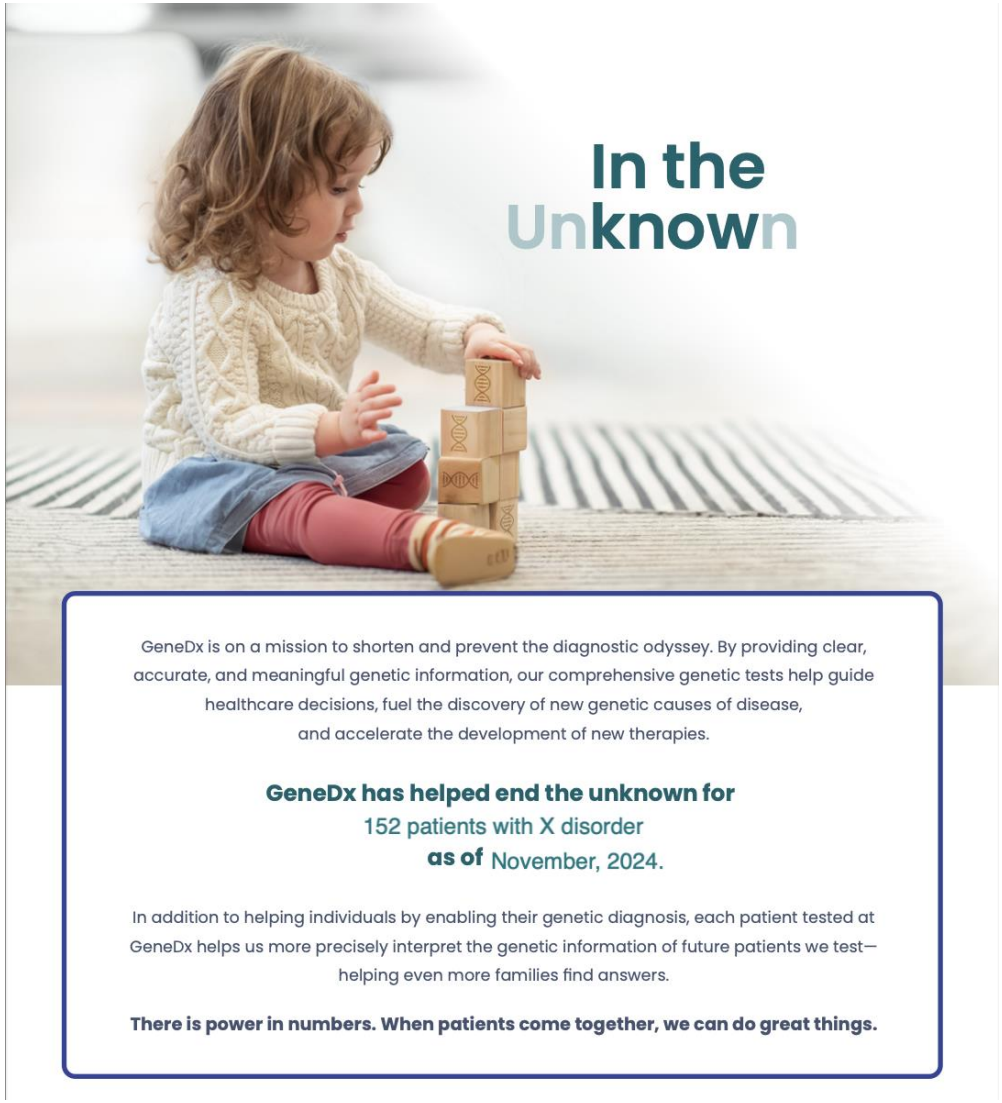


GeneDx can provide patient advocacy organizations with useful data.

Once per year, GeneDx will provide you with the number of patients that we have diagnosed with your disorder of interest.

This data can be helpful in understanding your community, as well as approaching potential biopharma partners.

Want to request this data for your organization? Email advocacy@genedx.com.



Want to grow your community? Be an advocate for genetic testing.

GeneDx

GeneDx's Patient Access Solutions help remove barriers to testing.

Epilepsy Partnership Program

The Epilepsy Partnership Program increases access to exome testing by offering a billing option if health insurance denies a claim or if you are uninsured. To learn more about this program, including who is eligible and how to apply, please visit **GeneDx.com/partnership-program** or scan the QR code.

Financial Assistance Program

GeneDx offers a robust Financial Assistance Program to help reduce the potential out-of-pocket costs associated with testing. For additional information, including a tool to estimate the level of assistance you may qualify for, visit **GeneDx.com/finanical-assistance-program** or scan the QR code.

A new path to access exome testing

Get connected with genetics experts, so you and your family can find answers.

Clinically impactful—and now easy to access.

Exome testing can help find the root cause of health concerns—especially for symptoms that can be caused by many different conditions. Yet for some children and their families, it can be difficult to find a local genetics expert to start the process.

GeneDx believes that everyone seeking a genetic diagnosis deserves answers. That's why we partnered with Genome Medical to connect families to telehealth genetic experts who can discuss exome testing and, if appropriate, order the test. Learn more at **GeneDx.com/genetics-connection** or use the QR code:

GeneDx/Genome Medical Partnership: How it works

1. Visit www.genedx.com/get-exome

2. Sign up and someone from Genome Medical will reach out to schedule a telehealth appointment, usually two or less weeks out

3. Discuss your child's medical history with a Genome Medical genetic counselor, who will then recommend the right test for you.

4. A cheek swab collection kit will be mailed directly to your family. After helping your child with the cheek swab, you will send it to GeneDx.

5. When the results are ready, a genetic counselor will notify you via email that you can download the results from their web portal.

6. You can discuss the results of your test with a Genome Medical genetic counselor and share results with your child's healthcare providers.

GeneDx is collecting patient stories. Please consider sharing yours with us.

GeneDx

Questions

Email us: advocacy@genedx.com
Visit us at: www.genedx.com