GENOME*XPRESS*

Rapid Genome Sequencing

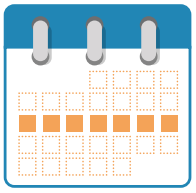
GeneDx
an OPKO Health Company

Genetic changes can cause over 13,000 different diseases and patients often present with overlapping symptoms.[1] Finding the correct diagnosis is not always straight-forward and may require multiple tests, costly evaluations, invasive procedures and long hospital stays.

Why Genome*Xpress*?

Genome sequencing (GS) analyzes the exons (coding DNA) in all genes and compares them to a database of reference sequences, similar to exome sequencing. Additionally, GS analyzes non-exome targets to find clinically relevant, non-coding variants such as those in promoter, intronic and untranslated regions. GS also provides copy number variant (CNV) detection for variants 1kb and larger.

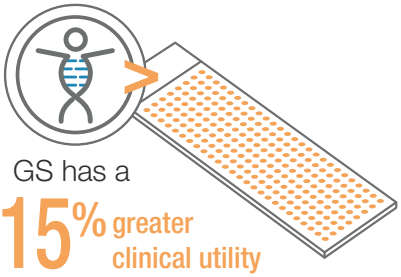
Genome*Xpress* is a rapid clinical genome sequencing (rGS) test that can find answers that more targeted genetic tests miss. It is especially useful when timing is critical and results may direct or alter medical management.



Verbal results in **7 days***

Speed to Diagnosis

- **Patients with genetic conditions can present with many complex issues. A rapid diagnosis can alter management, shorten the length of hospital stays, reduce healthcare costs and save lives.**
- **GenomeXpress provides a verbal result in 7 days* and a written report within 14 days*.**



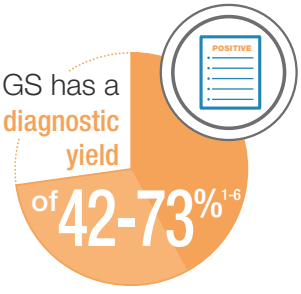
GS has a **15% greater clinical utility** than chromosomal microarray[2]

Clinical Utility

The proportion of patients in whom medical or surgical management was changed by diagnosis

Genome Sequencing:

- Has a more than 4× greater clinical utility than chromosomal microarray and should be considered a first-line genomics test for children with suspected genetic diseases.[2]
- Has been shown to demonstrate improved clinical outcomes, changes in medical management and net healthcare savings; particularly in acutely ill inpatient infants.[1,7]



GS has a **diagnostic yield of 42-73%**[1-6]

Diagnostic Utility

The rate of causative, pathogenic, or likely pathogenic genotypes in known disease genes

Genome Sequencing:

- Improves identification of disease-causing copy number and non-exonic regulatory and splicing variations allowing for identification of variants not detectable by exome sequencing.[6,8]
- Has been shown to successfully improve diagnostic rates for cases unsolved by exome sequencing and/or chromosomal microarray.[1,3,6,7]

**Reporting times are typical, but could be extended in situations outside GeneDx's reasonable control.*

207 PERRY PARKWAY GAITHERSBURG, MD 20877 • **T** 1 888 729 1206 (TOLL-FREE), 1 301 519 2100 • **F** 1 201 421 2010 • ZEBRAS@GENEDX.COM • WWW.GENEDX.COM

GENOME*XPRESS* IS THE ANSWER FOR MANY OF THE FOLLOWING INDIVIDUALS:

- NICU/PICU patients
- Patients seeing more than one specialist
- Those with unclear diagnoses
- Those with rapidly deteriorating health
- Those with developmental delay
- When multiple body systems are affected (e.g. multiple congenital anomalies)

Let Our Experts Help You

Order Genome*Xpress* from GeneDx and our experts will help you find the correct diagnosis for your patient. Our team includes hundreds of genetic counselors and MD/PhD scientists with extensive clinical experience and peer-reviewed publications. We go the extra mile to end your patient's diagnostic odyssey!

How to Order:

Email **Xpress@genedx.com** 7 days a week and a GeneDx Xpress Team member will assist with coordination of the testing process including:

- Facilitating completion of the test requisition
- Shipping sample collection kits to the provider/hospital for the patient and directly to parents
- Aiding in the consenting process
- Calling you with the verbal results 7 days* after all samples are received at our lab
- Supporting results interpretation

Test Information	
Turnaround Time	7 days* for verbal report 14 days* for written report
Verbal Results Delivery	Phone call to provider from Xpress team member; 7 days/week
Sample Type	Fresh blood in lavender top (EDTA) tube
Trios Required	No, but strongly recommended to increase diagnostic yield and to reduce the number of variants of uncertain significance (VUS)
Billing Options	Institutional or Self-Pay only

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References

1. Clark, et al. Sci. Transl. Med. 2019; 11, eaat6177
2. Clark et al. NPJ Genom Med. 2018; 3:16
3. Soden et al. Sci Transl Med. 2014; 6(265): 265ra168
4. Rady Children's Hospital-San Diego (2020). Project Baby Bear Final Report
5. Petrikin et al., Semin Perinatol. 2015 December; 39(8): 623 -631
6. Gilissen et al. Nature 2014; 17;511(7509):344-7
7. Farnaes et al. NPJ Genom Med. 2018; 3:10
8. Alfares et al. Genet Med. 2018 Nov;20(11):1328-1333

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