Newborn genomic research, delivered.



revvity

A comprehensive overview of Revvity's newborn sequencing offerings

Empower better newborn health from your laboratory.



For research use only. Not for use in diagnostic procedures.

Every sequence tells a story.

Newborn sequencing reveals the earliest genomic signals that shape health from the very start. Laboratory professionals now have an opportunity to lead this evolution – advancing the research that will redefine early detection and intervention for generations to come. The case for action has never been stronger.

30%

Children with rare diseases don't live to see their fifth birthday[1]



1 Hedrich, C. M. (2025). "Importance and Potential of Rare Disease Research in Pediatric Rheumatology and Beyond: Pushing Frontiers." ACR Open Rheumatology, 7(12), e70138. https://doi.org/10.1002/acr2.70138

4-5 yrs

Average delay until official diagnosis with rare disease[2]



2 Phillips, C., et al. (2024). "Time to Diagnosis for a Rare Disease: Managing Medical Uncertainty. A Qualitative Study." Orphanet Journal of Rare Diseases, 19(1), 297. https://doi.org/10.1186/s13023-024-03319-2

300M

Number of cases worldwide, a population equivalent to the entire United States[3]



3 Fehr, A., & Prütz, F. (2023). "Rare Diseases: A Challenge for Medicine and Public Health." Journal of Health Monitoring, 8(4), 3-6. https://doi.org/10.25646/11826

1-40+

Number of disorders that a typical screening panel might test for[4]



4 Scarpa, M., Bonham, J., Dionisi-Vici, C., et al. (2022). "Newborn Screening as a Fully Integrated System to Stimulate Equity in Neonatal Screening in Europe." The Lancet Regional Health – Europe, 13. https://doi.org/10.1016/j.lanepe.2022.100311

Revvity makes newborn sequencing research more accessible.

We help laboratories lead the charge. Leveraging our global genomic lab network, newborn screening heritage, and decades of experience in lab innovation, we bring your goals within reach. Whether you need an end-to-end sequencing workflow, expert-curated gene panel, interpretation software, or a custom solution, we're here to support your vision. See how you can partner with the global leader in newborn screening.

- **800M+** newborn samples analyzed
- **75+ years** of laboratory innovation
- **300M+** genOMIC tests performed*

*Tests performed by Revvity Omics laboratory network

Newborn sequencing workflows, reimagined.

Welcome to accessible sequencing. Covering every step – from DNA extraction of dried blood spot samples, to library preparation, automated liquid handling, sequencing, and final data analysis – this end-to-end workflow is built to make newborn research accessible to laboratories around the world.

One complete and configurable sequencing workflow



For research use only. Not for use in diagnostic procedures.

Workflow & lab services

Engineered for laboratory excellence.



Adaptable to your laboratory

Our modular workflow (with both manual and automated formats) offers multiple options for each step of the workflow, ensuring compatibility with your existing set up to support long-term success.



Empowering deep genomic research

Backed by an expertly-curated and continuously maintained panel of 390+ genes, complete with more than 100K annotated variants. The workflow can also accommodate whole genome sequencing (WGS) for broader exploration.



Field-tested and laboratory-proven

Deployed in population studies worldwide. Built by laboratory experts and bioinformaticians, it balances rigorous performance standards with practical usability for labs of all backgrounds.

Every component, from one partner.

Library preparation kits

Expert-maintained gene panel

Instrumentation

Genomic software suite

Your genomic research, your way.

We work with your laboratory to uncover the right NGS solution to achieve your goals.

1. Discuss laboratory direction & vision
2. Examine current lab capabilities & resources
3. Identify potential solutions

Flexible solutions for all laboratories.

Implementation type

In-house implementation
Your workflow, your way. With guidance from Revvity field application specialists

Lab-in-a-lab
Revvity managed installation. Ability to contract lab technicians for day-to-day workflow operations

Revvity Omics laboratory services
Outsource your research to Revvity's genomic experts

Genomic scope

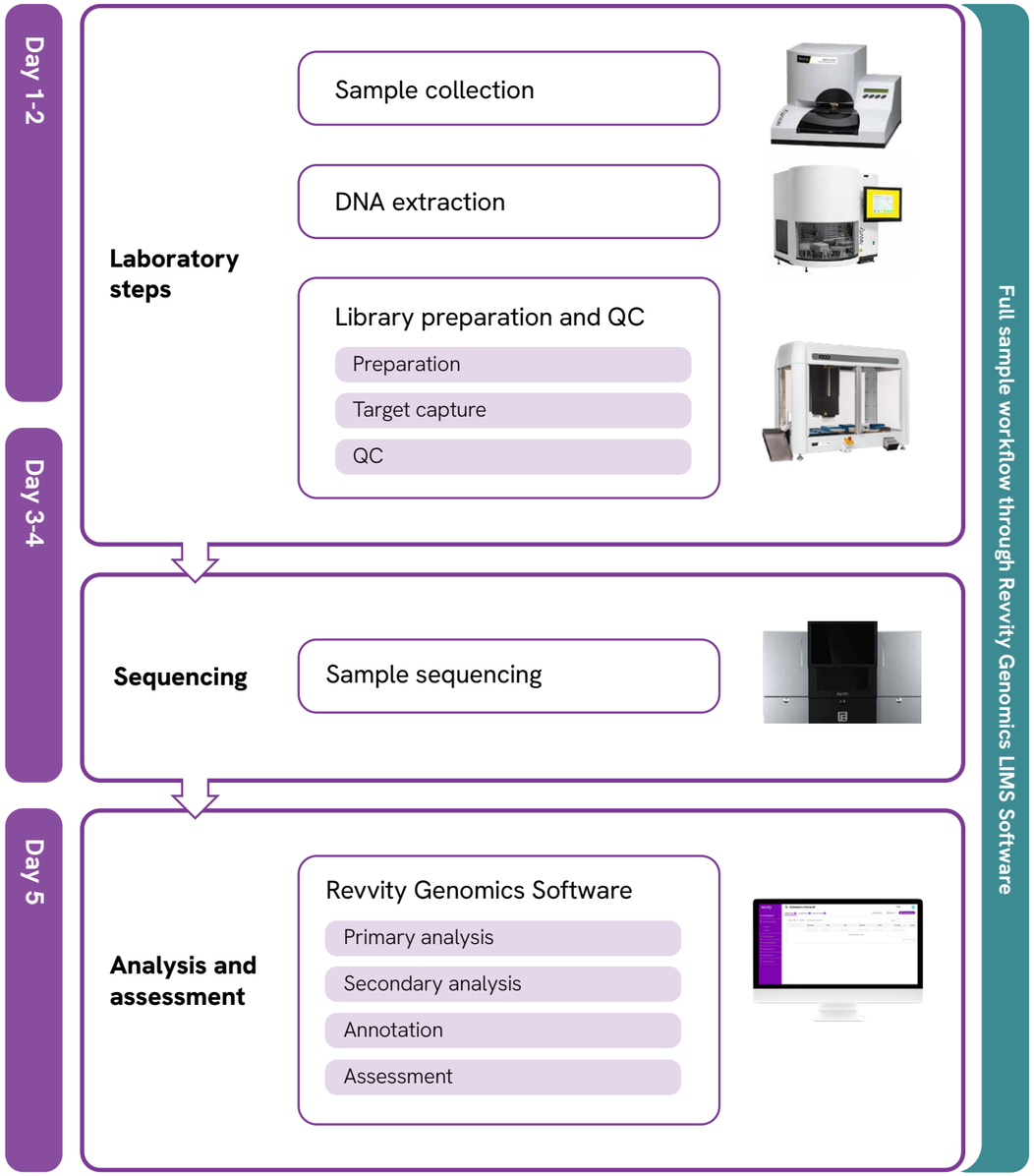
- Targeted gene panel
- Whole genome
- Whole exome*

* Whole exome only available for Revvity's laboratory service option.

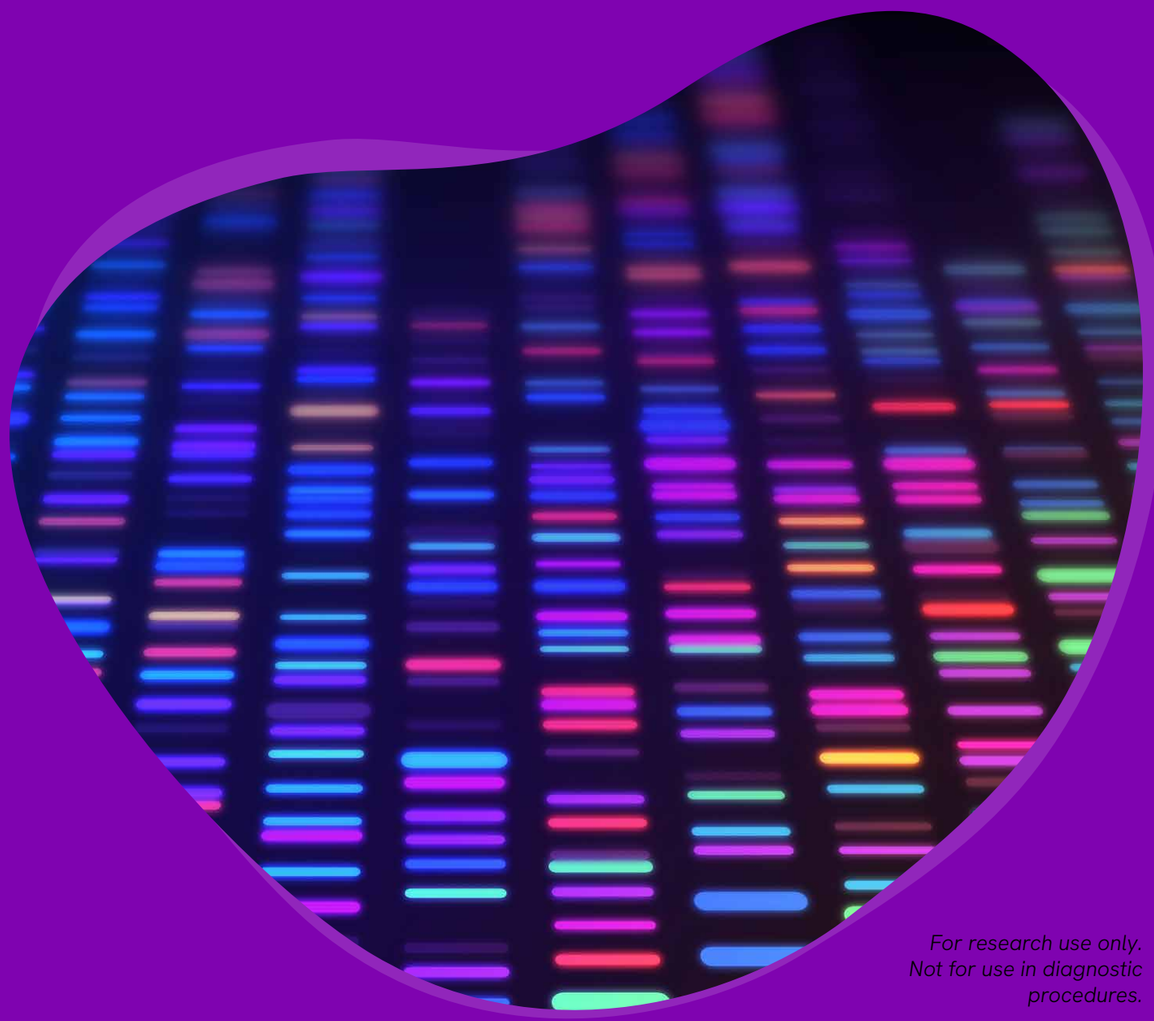
Sample type

- Dried blood spot (DBS)
- Whole blood
- Buccal swab
- Saliva

From sample to result in 4-5 days.



From sequencing to insight – in hours, not days.



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Software

Revvity Genomics Software.

Purpose-built for genomic workflows, our software suite is enhanced with lab-agnostic features that synchronize smoothly with your existing laboratory setup; bringing clarity and simplicity to even the most complex NGS (next-generation sequencing) operations.



Engineered by laboratory specialists

Accessible for labs of all backgrounds, designed for secure genomic analysis.



Primed for deep genomic insight

Carefully curated genetic panel of 390+ genes and 100K+ variants.



Configurable and adaptable for laboratories

A modular design that can adapt to multiple assays, external software and application areas.

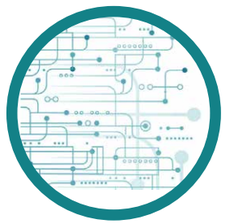
Key benefits.

Accelerated genomic analysis
Compresses interpretation from several days to less than 2 hours

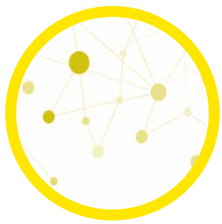
Simplified reporting
Reporting hub was designed by lab experts for organizations of all backgrounds

Uncompromising accuracy
Complete with state-of-the-art security and QC features for utmost reliability

One suite, three modules.



Revvity™ Genomics LIMS



Revvity™ Genomics Analyze

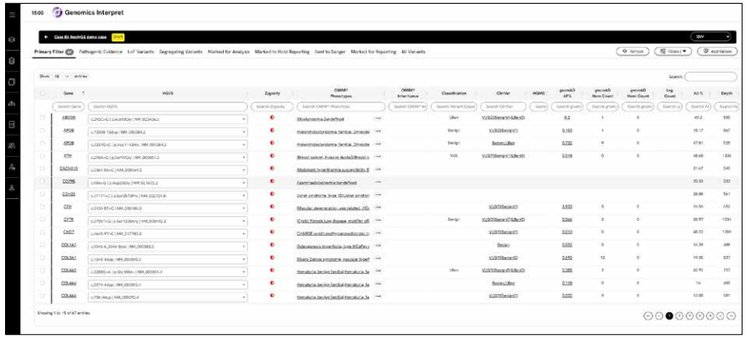


Revvity™ Genomics Interpret

Software

Accelerate your genomic analysis.

Cut down analysis timelines from 2-3 days to under 2 hours. Our software is equipped with powerful pre-filtering capabilities and pre-classifications, saving time without compromising performance. Features our gene panel of 390+ genes and a database of over 100K variants.



Revvity Genomics Interpret
Simplified filtering and pre-classifications for your cases

Reports that cater to your laboratory
A documentation hub designed by lab experts for organizations of all backgrounds. Present your findings with ease across a host of assay-specific templates.

Customizable set of templates to explore



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AnyPanel Sequencing and Deletion/Duplication Analysis

SAMPLE INFORMATION

Participant Name:	John Doe	**Specimen Type:**	DBS
Date of Birth:	08/21/2009	**Receive Date:**	08/08/2024
Sex:	Male	**Collection Date:**	08/06/2024
Accession ID:	2APP099	**Report Date:**	08/26/2024
Cross Reference:	Not provided		

RESEARCH PERFORMED

AnyPanel NGS Gene Sequencing and Deletion/Duplication Panel

No reportable sequence variants detected

RELEVANT FINDINGS AND INTERPRETATION

Copy Number Variants:

Classification	Event	Cytoband/Gene	Genomic Location	Size (bp)	OMIM
Benign	AOH	21q11.2 - q21.1	chr21:15,273,948-17,535,046	2261099	

21q11.2 - q21.1 AOH - Benign

NOTES

Genes and/or exons located in pseudogene regions are not covered in this assay. Additionally, mitochondrial genes are not included in this analysis.For each transcript listed, the analyzed region includes the coding exons and 10 bp of flanking intronic region on both sides of each exon. Select pathogenic deep intronic sites may also be targeted. In some cases, due to the complexity of the sequence, not all variants in the flanking intronic sequence are able to be analyzed. Variants are evaluated by their reported frequency in databases such as the Genome Aggregation Database (gnomAD), Human Gene Mutation Database (HGMD), ClinVar and other disease-specific or population-specific databases when applicable. Variants that have a population frequency greater than expected given the prevalence of the disease in the general population are considered to be benign variants. Pathogenic, likely pathogenic, and variants of uncertain significance are referenced. Benign and likely benign variants are not shown. Silent variants and intronic variants beyond +/-3 are not indicated unless known or suspected to be pathogenic. For those panels with 300 genes or more, variants are presented based on relevance to the research participant's information provided. For these large panels, there may be variants of uncertain significance present in the sample with partial overlap to some of the given phenotypic information which were determined not to be relevant enough for inclusion. The interpretation of variants is based on our current understanding of the genes on this panel. These interpretations may change over time as more information about the gene(s) comes available. A list of all variants identified in this individual is available upon request.

ISCN Nomenclature

21q11.2q21.1(15273948_17535046)x2 hmz

1

Software

Harmonize your informatics with one solution.

A modular software suite built to accommodate your lab.



Revvity Genomics LIMS

Create efficiency inside your lab.

Centralizes any lab workflow, connecting all steps seamlessly. Supports multi-assay analysis for enterprise-wide efficiency.



Revvity Genomics Analyze

Centralised hub for primary and secondary analysis.

Configure all analysis parameters for streamlined case management and categorization. Backed by a precise QC system ensuring maximum reliability.



Revvity Genomics Interpret

Deeper insight, actionable findings.

The crown jewel of Revvity's NGS software suite. Makes complex classification tasks more accessible to laboratories of all backgrounds.

State-of-the-art security and QC features.

Centralized security management
Our dedicated team monitors and maintains software safety and security

Secure cloud environment
AWS cloud and cloud operations are certified under the ISO/IEC 27001 information security management principles enabling HIPAA information security standards in-app

Configurable access control
Determine who can utilize the software based on role and responsibility

Data encryption
Encrypting data in transit (outside and inside the cloud), between each of the application components (in the cloud), and in the database and storage

Data Loss Prevention (DLP)
Preventing accidental exposure or infiltration of sensitive information, detecting and blocking the transmission of sensitive data outside the organization, or encrypting data before it leaves the cloud environment

Regular testing
External companies routinely audit our software's security to maintain robustness*

*Live customer data is not shared during these exercises

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Uncover the fast-track to genetic discovery.



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Gene panel

A rigorously curated panel for newborn genomics.

Our expertly-assembled 390+ gene panel captures the key genomic targets most critical to newborn research. Continuously refined by our global team of genomic specialists, it provides laboratories with a trusted, ready-to-use foundation for fast, confident interpretation.



Evidence-based panel

A carefully designed 390+ gene panel guided by newborn screening research, early-onset relevance, and global genomic evidence.



Extensive research applications

Our gene panel covers 17 different research fields, giving you the flexibility to study whatever genetic conditions interest you most.



Maintained by genomic experts

Continuously refined by Revvity's global team of genomic specialists to reflect emerging insights and best practices.

Accessible by design.

Genomic analysis shouldn't require deep bioinformatics expertise. Our panel and software suite provide a clear, structured framework that simplifies interpretation and enables laboratories of all experience levels to bring advanced sequencing research in-house. Trusted across our global lab network, it supports consistent, high-quality results worldwide.

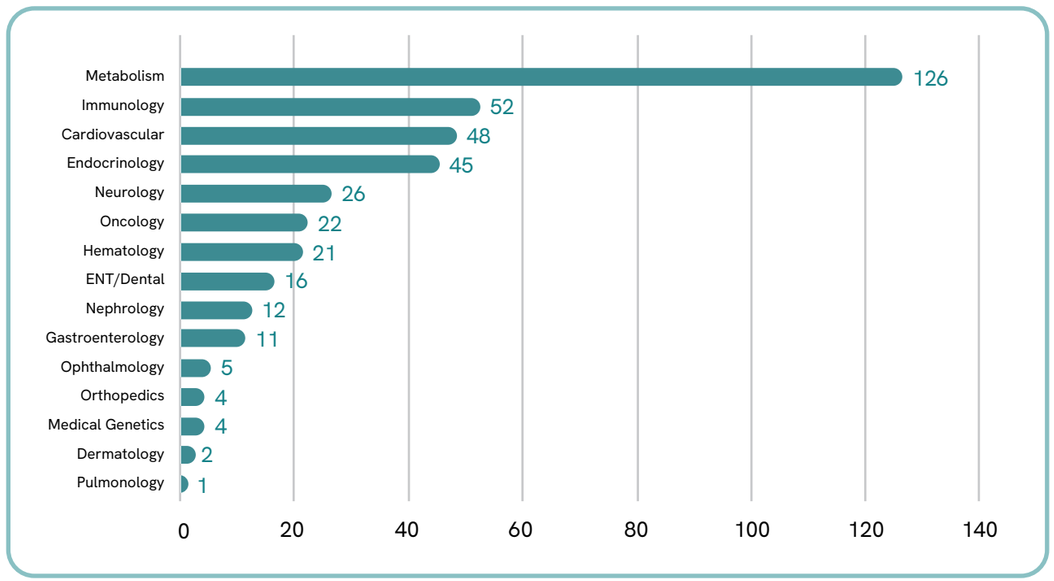
Comprehensive, evidence-based gene panel.

Backed by rigorous inclusion and exclusion criteria to ensure meaningful coverage of newborn conditions. Target selection reflects evidence from newborn screening frameworks and emerging research, taking into account:

- General recommendations (e.g. RUSP, pilot studies)
- Onset of the disease (< 2 years of age)
- Technical feasibility
- Prevalence and clinical relevance
- Availability of treatments or interventions

Extensive research applications.

Spanning 17 research areas and supported by continuously updated annotations, the panel enables laboratories to explore a wide spectrum of early-onset metabolic, endocrine, immunological, and genetic conditions with confidence and clarity.



Maintained by genomic experts.

The panel is curated and refined by Revvity Omics' multidisciplinary clinical team, including:

- Genomic experts
- Bioinformaticians
- Genomic analysts
- Board-certified directors
- Genetic counselors
- Laboratory scientists

Targeted panel or WGS: which is right for you?

Our workflow supports both targeted sequencing and whole-genome approaches. For most laboratories, we recommend starting with targeted NGS due to its reduced complexity, smaller data footprint, and more streamlined interpretation requirements. Connect with our team to determine the best fit for your laboratory and research goals.

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The Revvity difference.

Partnering with Revvity means gaining more than advanced technology. You gain a team of genomic experts and global support specialists committed to your success. We provide the tools, insight, and hands-on guidance needed to drive your newborn sequencing program forward with confidence and efficiency.

Accelerate your science with Revvity today.

Empowering academic & translational research across

190

countries

~45% Americas

~30% Europe

~25% Asia



Lead the charge in newborn genomic research.

In-house implementation

Your workflow, your way. With guidance from Revvity field application specialists.

Learn more

Revvity Omics laboratory services

Outsource your research to Revvity's genomic experts.

Learn more

www.revvity.com

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