WHITE PAPER

Redefining the genomics landscape: Navigating the future of personalized medicine

Executive summary

"Genomics" is the branch of science concerning the study of all the DNA of an organism (its "genome"). One benefit of studying this information is that it allows for a greater ability to predict, identify, and treat diseases with greater precision for each individual.

Initiated by genomic testing, genomic medicine is now firmly established as a vital component of a modern healthcare system. With 80% of rare diseases having a genetic basis[1], genomic testing can reduce both the time and number of hospital visits required to arrive at a diagnosis, improving both patient experience, patient health outcomes and total cost to the healthcare system. Genomics also plays a part in common disease like asthma where genomic knowledge and information can assist healthcare professional to better monitor and ultimately treat symptoms.

Genomic testing can help reduce post-diagnosis costs by suggesting which treatments are most likely to have a positive, negative, or neutral outcome for individual patients. Today, CRISPR-based cell manipulation techniques and payload delivery systems, such as the use of adeno-associated virus (AAV), provide the ability to target and correct defects in the genetic code in cells. Current efforts in a common neuromuscular disease like Duchenne Muscular Dystrophy (DMD) use this approach to provide personalized therapy based upon the individual's genomic cause of DMD. Gene therapy has shown promising results in the treatment of hereditary ocular diseases such as retinitis pigmentosa and Usher syndrome. It also allows clinicians to identify the most effective treatment options quickly and effectively, enabling patients to receive personalized treatment as quickly as possible. Despite the significant benefits genomics offers to patients and healthcare systems, successful implementation of a genomic program requires a great deal of time and expertise.

Revvity is a global life science and diagnostic organisation with a mission to accelerate the therapeutic process from discovery to cure. Revvity can do so by simplifying the complex challenges associated with genomic program setup, and by providing labs with a complete, single source answer. We collaborate with interested organizations to understand their testing needs and thus provide a bespoke solution that is specifically catered to those needs. Solutions encompass every aspect, including laboratory space build out, sample collection, clinical grade testing workflow, training, management of local talent, and generating a final clinical grade report.

Revvity's workflow solutions are highly customizable to the specific needs of each unique lab and can scale in throughput using pre-tested automation. Revvity's testing areas of expertise include, but are not limited to:

- Pre-conception screening such as carrier screening and prenatal testing
- Post-natal testing including:
  - Newborn Screening (NBS) and Sequencing.
  - Genome sequencing with application in rare disease

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  - Panel testing in areas such as Hereditary Cancer, Cardiovascular, Neuromuscular among others.
- Other testing in areas such as Infectious diseases and Immunology

The challenge

Finding a way to implement a genomic testing program within a laboratory or healthcare system can be a complex endeavour with multiple barriers that need to be acknowledged and addressed:

- The field of genomics is rapidly evolving, with new technologies, methodologies, and discoveries emerging constantly
- Genomic testing involves sophisticated techniques, equipment, and analyses
- Proper hiring, training, and retaining of skilled personnel with the required expertise
- Developing and maintaining high quality standards and accreditation requirements (essential to obtaining accurate and reliable results)
- Knowing how to navigate the complex regulatory frameworks and stringent regulatory requirements
- Ensuring robust data management systems are implemented to securely store, analyze, and interpret genomic data while safeguarding patient privacy and compliance with data protection regulations (such as GDPR and HIPAA)

The solution – genOMICS labs

To easily overcome these barriers, Revvity developed innovative, comprehensive, and customizable solutions to fast, reliable, and flexible genomic testing services that keep pace with cutting edge technological and scientific developments. Revvity operates a global network of state-of-the-art genomic testing laboratories staffed by experienced clinical geneticists. By offering genomics testing as a service, Revvity can provide governments, clinicians, and research institutions the opportunity to rapidly realize the benefits of this technology, with a diagnostic menu tailored to both national and local requirements.

With laboratories in China, India, Sweden, the United States and the United Kingdom, our laboratory network helps cater to the world for its genOMICS testing needs. Given our experience of operating in different regions, we understand the population specific requirements and work with both an in-region for-region and in-country for-country approach.

We also provide a “lab-in-lab” solution that transfers the testing service to an in-house model, run by the client, as part of the overall service plan. This flexible option enables organizations to rapidly access the most cutting-edge genomic applications with minimal capital expenditure, while planning to bring testing in-house, if desired. We are committed to ensuring our partners are included in service delivery, successful transfer of knowledge through fully immersive training, updates on technological advances, and full program management from outsourced services to in-house delivery.

The benefits

This flexible approach to genomic testing offered through Revvity can lead to many benefits, including cost savings, expedited time to diagnosis, and improving patient outcomes through personalized treatment plans.

Various studies have demonstrated that the incorporation of genomic testing services into healthcare systems can not only shorten the time to diagnosis for a patient but can also generate substantial cost savings for healthcare systems by allowing medical treatments to be used in a more targeted and effective way. Examples of these studies include “Project Baby Bear” conducted in California, USA which found that among the 178 babies who had genomic sequencing performed, there was an average of 513 fewer days spent in the hospital, 11 fewer major surgeries, and an estimated $2.5 million in healthcare savings realised[22].

Revvity Omics laboratories

Providing biochemical screening and clinical OMICS services in support of pharma and biotech clinical programs and new assay development services

Global testing capabilities

Pittsburgh, PA
30,000 sq ft
- Prenatal
- NBS
- NGS
- Molecular Genetics
- Cytogenetics
- Biochemical Genetics
- NIPT
- Immunology

Atlanta, GA
4,000 sq ft
- Center of excellence

Hebron, KY
32,000 sq ft
- ViaCord Cord Blood & Tissue
- Stem Cell Biobank for Omics Interpretation

Cambridge, UK
30,825 sq ft
- Preclinical services
- Mimix reference standards
- Bioproduction
- Base editing

Manchester, UK
1,500 sq ft
- NGS

Oxford, UK
41,516 sq ft
- T & B cell Immunoassay

Sollentuna, Sweden
1,000 sq ft
- Maternal Health
- NIPT
- NGS

Chennai, India
14,000 sq ft
- Prenatal
- NBS
- NGS
- Molecular Genetics
- Cytogenetics
- NIPT

Taicang, China
17,000 sq ft
- Prenatal
- NBS
- NGS
- Molecular Genetics
- Cytogenetics
- Biochemical Genetic
- NIPT
- PGD

- State-of-the-art, clinical grade **global laboratory network**
- **Deep expertise** in multiple omics platforms with demonstrated implementation in newborn screening, clinical omics, biochemical and immunology services, and new assay development
- **Clinical staff** including board-certified geneticists, genetic counselors and laboratory scientists

Disclaimer: This testing service has not been cleared or approved by the U.S. Food and Drug Administration. Testing services may not be licensed in accordance with the laws in all countries. Laboratory licensing and accreditation, as well as the availability of specific test offerings, is dependent upon laboratory location. The content of this pamphlet is provided for informational purposes only, not as medical advice. It is not intended to substitute the consultation, diagnosis, and/or treatment provided by a qualified licensed physician or other medical professional.

Introduction

The completion of the Human Genome Project has opened the floodgates for the betterment of human health. Genomics has revolutionized our ability to save lives, lower medical costs, and improve outcomes. We have seen this through the improvement of disease diagnosis, the development of vaccination and public health initiatives, and the personalization of effective treatments through proper understanding of the underpinning of an individual's condition, be it cancer, genetic disease or choosing the most effective medication.

Burden of rare disease

There are approximately 7,000 rare diseases that have been described: 80% have genetic origins, with 70% starting in childhood.[1] These illnesses affect an estimated 30 million people across the US, and approximately 300 million people worldwide.2 The journey to the correct diagnosis in a higher income country may take an average of 4 to 5 years with visits to multiple specialists and entail several misdiagnoses.[3] The long road to diagnosing these patients presents one of the most confounding challenges affecting the survival and well-being of these individuals and their families.

Rare diseases are responsible for 35% of deaths in the first year of life, and because of the high prevalence in childhood, 30% will not live to see their 5th birthday.[4]

In addition to being devastating and traumatic for the patients and families, the misdiagnosis of rare diseases often leads to patients experiencing significant health consequences such as high morbidity and mortality, psychosocial and occupational burden, and substantial financial and medical burden on both the family and the overall healthcare system. The economic burden of rare diseases includes both direct (medical and nonmedical) and indirect costs. Direct medical or healthcare cost burden includes hospitalizations and emergency visits. More than 10% of inpatient costs are from patients with rare diseases.[5]

A recent retrospective study of medical and insurance records indicates that healthcare costs for people with rare diseases have been underestimated. Costs are three to five times greater for those with a rare disease than those without one. The study, led by the National Institutes of Health's National Center for Advancing Translational Sciences (NCATS), provides evidence of the potential impact of rare diseases on public health, suggesting nationwide medical costs for individuals with rare diseases are on par with those for cancer and heart failure.[6]

One study indicated that the cost for an undiagnosed rare disease patient over ten years was 100% more than the average patient due to more outpatient visits. This cost differential was heightened in pediatric patients.[7]

Burden of undiagnosed disease

As many doctors and specialists are unfamiliar with the symptoms of syndromes and rare diseases, it can often lead to patients being misdiagnosed. Frequently these patients exhibit symptoms similar to more common diseases or are nebulous, making the diagnosis even more complicated and consequently leaving patients and families frustrated as they continue their diagnostic journey.[8]

Getting an accurate diagnosis is the first step towards finding the proper treatment. Yet even before treatment, an accurate diagnosis is what patients most desire for their peace of mind. Numerous patient testimonials exist in the literature about the extreme difficulties of receiving an accurate diagnosis. The norm is a long, frustrating process that delays proper clinical care and potential treatments.[9,10]

These stories around misdiagnosis and underdiagnosis are not unique. In one study, rare disease patients consulted with nearly eight physicians before receiving an accurate diagnosis and up to 23.5% of respondents had seen between 6-10 physicians.[11]

Consequences of delayed or missed diagnoses

Most patients suffering from rare diseases or inherited conditions only receive symptomatic therapy. Many patients experience barriers in access to care, and fewer than 10% receive disease-specific treatment.[7] Even brief delays in diagnosis may have profound effects on outcomes; for over 40% of rare disease patients, treatment delays are precipitated by misdiagnoses.[8]

For inherited conditions, knowing the causative variant and the mode of inheritance informs the clinician, patient, and family on the best clinical care; educates the patients about the potential of passing the disease onto future generations; and, if desired, evaluates alternative family planning options. However, as discussed, the diagnostic delay or misdiagnosis varies from months to decades, depending on the patient's phenotype, age, and available resources. On average, rare disease patients wait 4 to 5 years to receive a diagnosis[3], with some waiting over 20 years.[12] The consequences of delayed or missed diagnoses lead these individuals down a path where they face a diagnostic odyssey and continue to undergo extensive and expensive workups at several institutions.[13] Rare diseases burden families and the US healthcare system:[14]

- 28% of neonatal intensive care deaths are caused by rare diseases.
- 65% of rare diseases are associated with a shortened lifespan.
- 3-10% of hospitalizations (regardless of age) are related to a rare disease[4].

Without intervention, an individual's health can significantly deteriorate as the rare disease progresses, and people may receive inappropriate care, have lower expectations in life, and experience a loss of hope.[12]

Eighty percent of rare diseases have genetic origins[1], which lays the groundwork for the critical role of genetic and genomic testing as a logical tool that can assist clinicians in finding a clinical diagnosis that may ultimately lead to changes in care, combatting these illnesses, ending the diagnostic odyssey, and improving outcomes for the patients and families.

Genomic testing

Identifying an infant, child, adolescent, or adult with a rare genetic condition or inherited disease may include several targeted strategies. Historically, the diagnostic strategy often integrates several specialists that contribute to the overall clinical workup and includes a combination of several sub-specialties that work in tandem due to the rarity and clinical phenotype of these patients. This strategy often includes diagnostics from the radiographic, biochemical, electrophysiologic, and genetic testing disciplines leading to a high burden on the patient and the healthcare system.[15,16]

Genetic and genomic testing strategies play a pivotal role in this diagnostic strategy. Such testing is often life-changing to the patient, as it may end their complicated odyssey to finding a correct diagnosis and ultimately provide comprehensive answers, and possibly treatments, sooner.[16]

Molecularly confirmed diagnoses are pivotal, as they often lead to improved healthcare outcomes ranging from the use of specific enzyme replacement therapies; avoiding surgical interventions associated with misdiagnoses; enhancing knowledge and surveillance for unexpected comorbidities associated with certain rare disorders; and reducing the financial and psychological impact on the patients, family, and healthcare system. They can also inform relatives, including siblings and parents, of potential prenatal diagnostic options.[16]

While various genetic tools include targeted approaches and broad genomic sequencing, whole exome (WES) and genome sequencing (WGS) are diagnostic strategies that play a crucial step in tracking down a diagnosis and evaluating genetic conditions with high degrees of heterogeneity.[15]

The rationale for investment in genomics

Genomic testing is now firmly established as a vital component of a modern healthcare system. With 80% of rare diseases having a genetic basis[1], genomic testing can reduce both the time and number of hospital visits required to arrive at a diagnosis,

improving both patient experience, patient health outcomes and total cost to the healthcare system.

Although the applications of genomics in healthcare are growing daily, there are a number of challenges associated with embedding genomics into the clinical offering, including infrastructural, technological, educational, and political constraints. This complexity sometimes makes healthcare systems and governments pause their policies, thereby preventing patients and healthcare systems from reaping the benefits.

Additionally, hospital budgets are not currently set up to enable the rapid incorporation of new and quickly evolving technologies into clinical lab settings, with specialist knowledge required to both set up and deliver services, along with significant up-front expenditure on property, plant, and equipment. These investments require constant re-investment to remain innovative and relevant in a rapidly moving sector. As healthcare systems are often unable to build an in-house system from scratch, many find themselves searching for an optimal solution to help their patients.

Revvity's Omics' areas of expertise

Revvity Omics can offer a shortcut to fast, reliable, and flexible high complexity multi-OMICS testing services that keep pace with technological and scientific developments. Revvity operates a global network of strategically placed state-of-the-art genOMICS testing laboratories staffed by experienced clinically trained team members. Having this network that continues to drive forward not only globally recognized patient testing services but significant levels of R&D, allows customers to benefit from shared learning and a collaborative community.

Through offering genomics testing as a service, Revvity can offer an opportunity to rapidly realize the benefits of this technology, with a diagnostic menu tailored to both national and local requirements. As part of Revvity's offering, we are uniquely able to transfer the high complexity testing workflow to an in-house model, run by the client, as part of the overall delivery plan.

This approach would enable a system to rapidly access the newest Omics applications, with limited capital outlay, whilst planning to bring them in-house in a timeframe that suits the budget and strategic priorities of the organization.

This flexible approach to genomic testing offered through Revvity and Revvity Omics may bring many benefits, including cost saving through speeding up the time to diagnosis and improving patient outcomes through personalizing treatment plans.

Numerous studies have shown that the thoughtful incorporation of genomic testing services into healthcare systems can not only shorten the time to diagnosis for a patient but also generate substantial cost saving for healthcare systems by allowing medicines to be used in a more targeted and effective way and freeing up hospital beds quicker. Examples of these studies include "Project Baby Bear" run in California, USA (United States of America) which found that among the 178 babies sequenced, there was an average of 513 fewer days in the hospital, 11 fewer major surgeries and an estimated $2.5 million in healthcare savings realized[22].

A quick peek at Revvity

- Built on 75+ years of laboratory expertise
- Over 70 newborns are saved every day thanks to our technology
- Serving 190 countries
- 40 million babies are screened annually for life threatening diseases using our technology

Major potential benefits

- Provides accelerated access to cutting edge genomic technologies, expertise, and service capabilities.
- Reducing time to diagnosis frees up hospital beds, enabling the ability to treat more patients and improves patient outcomes.
- Personalized medicine allows expensive medicines to be used in a more targeted manner, reducing costs.
- Unique Revvity service transfer model allows groups

to plan when to bring services in house and work with Revvity Omics to ensure services transition smoothly.

- This model allows for greater flexibility when planning budgets, enabling large capital expenditures to be planned and defined well in advance.
- Working in partnership, full transparency of operational costs is shared from the outset. This allows for a full cost-benefit analysis when evaluating which services it brings in-house and helps to facilitate future planning.
- For services that are bought in-house, full immersive training in the workflow (at a Revvity Omics laboratory or on-site) is available. Enabling technicians to be fully up to speed with the process before it is even launched in-house.
- Ability to design, develop, manufacture, validate, and implement assays specific to customer's needs.
- Fast tracks the delivery of the government's strategic goals.
- Upskilling local talent with the know-how of how to perform high complexity testing using cutting edge technologies.

In the global race to realize these benefits, specialized resources are difficult to both find and develop. At Revvity, we are committed to ensuring our partners are included in service delivery. When time appropriate, Revvity will look to transfer knowledge through fully immersive training and enable local teams to seamlessly take over operations and transition to a fully in-house delivery model.

Collaboration showcase

Accelerating the deployment of clinical grade testing

Although our laboratories are set up to provide clinical testing and provide services to hospitals and clinics, we also have the experience and expertise to support the set-up and scale-up of external laboratories.

Our turnkey solutions for clinical testing offer a complete spectrum of screening and diagnostic technologies that enable the early clinical insights required for a proactive approach to healthcare. Our "Lab-in-Lab" concept offers clients a complete package of on-site assay validation and automation, technical and operational leadership, sample collection packs, quality management, lab information, and reporting support. Clients only need to provide the space for setup. We operate approximately nine such Lab-in-Labs in India and have similar set-ups in the USA.

One recent successful example is the rapid setup of COVID testing laboratories in Cardiff, Wales; Los Angeles, California; and Midlands, England which were designed to process up to 85k COVID samples per day capacity at launch, with scalability of up to 220k samples per day at full capacity.

Expediting the process from drug discoveries to therapeutics

With our instruments and services, Revvity can support non-profits, government institutions, research organizations and pharma throughout their therapeutic development journey.

	Goals	Revvity solutions
STEP 1 **Discovery & development**	1. Target identification 2. Assay development	▪ Assay development for biomarker identification. ▪ Cell line engineering services for generation of cell models. ▪ CRISPR, RNAi, base editing, and gene expression platforms for determining gene function and target ID. ▪ Analysis solutions to reliably detect, analyze and characterize cells, protein or nucleic acids.
STEP 2 **Preclinical research**	1. Authentication of identified target 2. Lead development and optimization 3. *In silico* assay screening	▪ Combine functional genomic and cell panel screening services to identify and confirm targets. ▪ Test and optimize different formulations or oligos using high-throughput platforms. ▪ Functional genomic screening services to identify drug mechanisms. ▪ Immune response and toxicology insights with immune cell assay services.
STEP 3 **Clinical development**	1. Sample collection, storage & logistics 2. Individual stratification 3. Testing for clinical trials	▪ Providing end-to-end support required for sample collection to clinical testing solutions using next generation sequencing technologies. ▪ State-of-the-art bioinformatics pipelines designed to deliver the most reliable data processing and interpretation. ▪ Fluorospot and Flow cytometry to measure the efficacy of your vaccine or immunotherapy. ▪ Individual stratification for clinical trials through our global laboratory network.
STEP 4 **FDA review**	1. Regulatory compliance 2. FDA approval application	▪ Guidance for regulatory and compliance approval. ▪ Through a dynamic process, we consult with regulatory bodies to facilitate the introduction of innovative products to market.
STEP 5 **Companion Diagnostics**	1. Manufacturing and commercialization of CDx kits	▪ Ability to develop and commercialize the kits for companion diagnostic assays for targeted therapies.
STEP 6 **Study expansion**	1. Program management 2. Global laboratory network	▪ Dedicate program management support through out the lifespan of the study. ▪ Global commercial footprint for deeper reach. ▪ Support to scale up the study through the global lab network.

Spotlight: The benefits of newborn screening and sequencing

Newborn screening (NBS) plays a vital role in both saving and improving babies' lives. It has been shown in emerging economies that addressing child health issues creates a positive impact[27]. Newborn screening could further help improve infant and child mortality rates.

It seems particularly noteworthy that, as the infant mortality rate (IMR) decreases, newborn screening has gained traction as an emerging healthcare strategy worthy of funding consideration. A 2015 report notes that countries within the Asia-Pacific region whose IMR was 7 per 1000 births have been able to achieve at least 90% NBS coverage[25].

Hundreds of thousands of babies each year worldwide could avoid a lifetime of disability or even death. Unfortunately, not all families and their babies have access to the wide range of available tests. Of the 134 million babies born in the world each year, only about one third receive screening of any type, and many babies are only screened for one or two conditions.

Next-generation sequencing (NGS) has revolutionized newborn screening by offering several benefits over traditional screening methods. NGS allows for early, simultaneous screening of multiple genetic disorders in a single test, enabling the detection of a wider range of conditions while reducing the need for multiple tests, timely intervention and treatment which can prevent or mitigate the onset of symptoms, improved health outcomes, and reducing the burden of disease on patients and families. Its high sensitivity and specificity minimize the likelihood of false-positive or false-negative results, thereby providing more accurate and reliable screening outcomes. Faster turnaround times for screening results enable more timely interventions and clinical management. NGS platforms are highly flexible and scalable, allowing for customization of screening panels based on specific populations, geographic regions, or clinical indications. This flexibility enables healthcare providers to adapt screening protocols to evolving knowledge of genetic diseases and emerging public health needs. NGS is paving the way for personalized medicine approaches tailored to each patient's unique genetic profile.

Case study examples

Revvity's clinical testing services offer a range of biochemical and genomic analyses to reliably diagnose rare disorders, especially in newborns, aiding early intervention and management. Our lab-in-lab concept enables Revvity's laboratories to function as a backup for processing client laboratory samples while maintaining the highest level of quality and data security.

Case study 1: Ultrarapid whole genome sequencing

A 3-day old male child presented to his physician with hypotonia, significant birth defects and metabolic acidosis. The physician ordered our Ultrarapid whole genome sequencing in conjunction with our biochemical screening. Within 53 hours from receiving the sample in the lab, the testing revealed elevated levels of Propionylcarnitine (C3) on the biochemical profile and identified a pathogenic sequence variant in the *PDHA1* gene.

The fast turn-around time allowed for an early detection and diagnosis in this child, which provided the healthcare providers with the ability to start with early intervention strategies in order to give this child the best possibility of an improved outcome. This chance would not have been possible without our lab's quick turnaround time and comprehensive genetic analysis.

Case study 2: Backup support for state-run labs

Oftentimes, laboratories may need assistance in running their tests. Staffing, sample volume, lab remodeling or relocation, new technology transfers and legislative mandates may all put a temporary pause to the laboratory's ability to function smoothly. Revvity has laboratories in place to help with these hiccups and ensure the proper and seamless operation of a newborn screening program. Revvity provides emergency newborn screening backup services to several of States in the United States in support of State newborn screening programs.

Case Study 3: Lab-in-Lab Concept

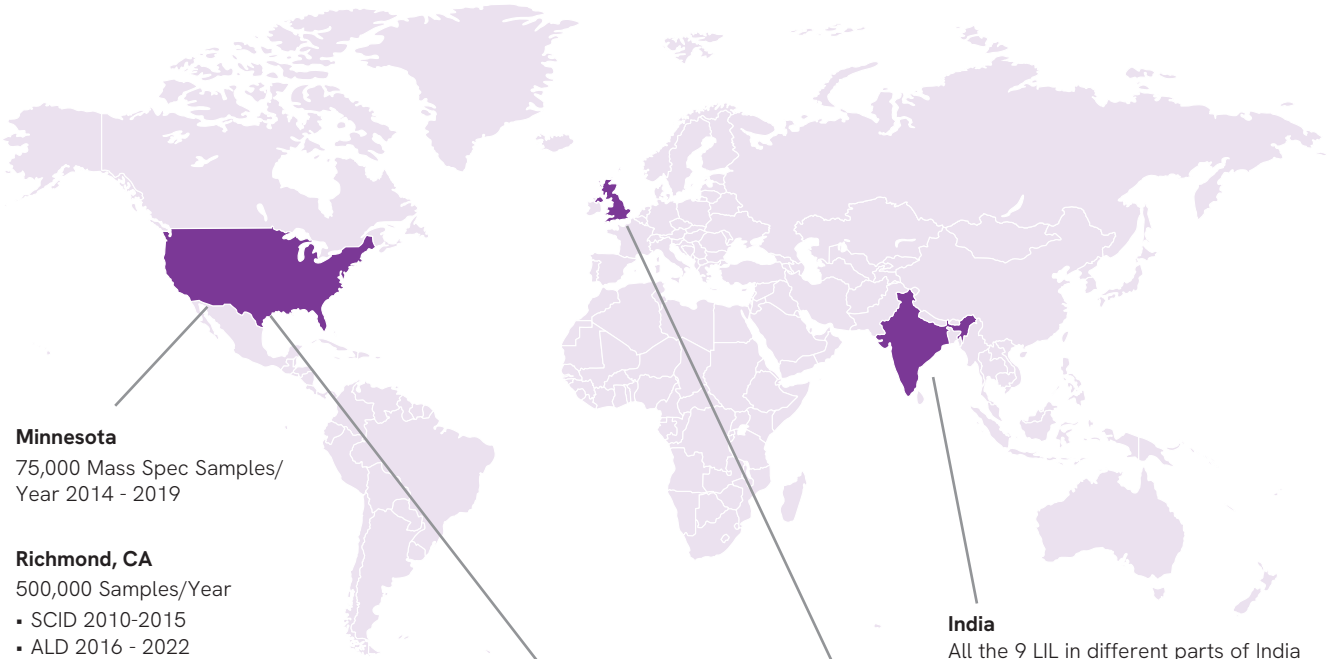
Occasionally, laboratories are not ready to run a newborn screening system by themselves. This could

often be due to lack of expertise, infrastructure and/or financials. To provide a bridge in the transition to running a newborn screening program themselves, many laboratories have asked Revvity to place a smaller version of our own laboratory within their laboratory. This allows laboratories to learn, evaluate, and align their needs, without the commitment of the day-to-day running of the systems. For example:

- Lab-in-Lab (LiL) contract with North Carolina where they provided us with space to set up instruments and run the testing
- Multiple LiLs in India that process prenatal biochemical testing and newborn screening

These case studies demonstrate how Revvity's comprehensive biochemical and genetic analysis, quick turnaround time, and ability to process large amounts of samples can aid in early intervention and management of genetic disorders in newborns. Our lab-in-lab concept also offers complete data security and provides valuable backup support for client-run labs in the US.

Lab-in-a-lab case examples



Minnesota
75,000 Mass Spec Samples/ Year 2014 - 2019

Richmond, CA
500,000 Samples/Year
- SCID 2010-2015
- ALD 2016 - 2022

Valencia, CA
CDPH Branch Laboratory in Valencia, CA
- Designed to process up to 40K COVID19 samples at launch with scalability up to 150K samples at full capacity
- Operational laboratory built in 8 weeks from contract completion

Specifications
- 135,000+ square foot lab
- 55 Revvity instruments installed at launch increased up to 158 instruments at full capacity
- 400 new Revvity employees added at launch increasing to approximately 1200 employees at full capacity

North Carolina
130,000 NeoBase2 Samples/ Year 2021-Present

Florida
250,000 Samples/Year
- SCID 2012-2019
- ALD 2018-2019
- SCID/SMA 2020

India
All the 9 LIL in different parts of India has been set up to process Prenatal Biochemistry samples

Newport, Wales & Charnwood, England
Lighthouse Laboratories
- Designed to process up to 60K COVID19 samples at launch with scalability up to 70K samples at full capacity
- Operational laboratory in 4 weeks from site handover

Specifications
- 15,000+ square foot lab
- 22 Revvity Instruments installed at launch
- 600 new employees at launch

Conclusion

Approximately 30 million people in the United States[2] and 300 million worldwide are affected by one of the 7,000 rare diseases[26]. 25% of pediatric hospitalizations are due to one of these conditions and often will lead to significant economic burdens for patients, families, and the healthcare system. Unfortunately, the medical diagnosis of a rare disease is challenging to achieve by clinical work-up alone. As a result, the patients and families undergo a diagnostic odyssey that includes multiple evaluations, specialists, hospital admissions, therapies and studies that can take anywhere from 5 to 30 years until they receive an accurate diagnosis. It is therefore not surprising that these rare diseases often lead to early morbidity and mortality.[3]

The use of genomics in clinical practice is advancing rapidly and holds great promise for patients and families in identifying patients at risk. As discussed, in critically ill patients, rapid genetic diagnosis has demonstrated improved clinical management, overall prognosis, and a reduction in costs. Recent programs in neonatal populations have reduced turnaround time to days and have shown that rapid genetic diagnosis enhances patient care and reduces healthcare costs.

In the last seven years, we have actualized many of the benefits of utilizing genomics in clinical practice, including:

- The diagnosis of numerous novel genetic conditions[17]
- A reduction in the costs and psychological burden to families and the healthcare system[4,9]
- Earlier detection of conditions for better pregnancy management, including prenatal diagnostics[18,19]
- Assessing proper risk by screening patients and families with hereditary cancer risks[20]
- Fast results by using newborn screening for infants in the NICU[21,22,23,24]

Genomics has the potential to revolutionize medicine through a personalized approach, answering long-awaited questions, such as what disease do I have? How was it caused? Will it eventually progress? Is there a promising treatment? And ultimately, will other family members be affected? These are all critical questions for the provider, patients, and family members as the first step toward the medical management of genetic diseases.

Genomics is significantly changing clinical practice and care and demonstrating proven results of ending the diagnostic odyssey, otherwise known as that long road to the "final diagnosis," by way of achieving a higher diagnostic yield, delivering a faster time to diagnosis, informing treatment decisions, avoiding incorrect treatment or therapy, lowering overall costs, improving quality of life, and allowing providers a quicker window of opportunity to create long-term treatment plans.

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