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# Transforming Care, Saving Lives

## Revitalising UK Life Sciences and Diagnostics

December 2024

revvity

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# Innovative solutions for Life Sciences and Diagnostics

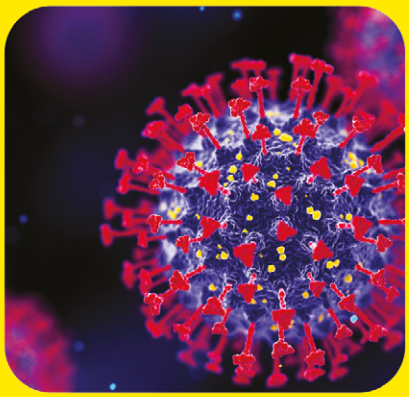
Maternal Fetal Health

Newborn Screening

Drug Discovery

Next Generation Sequencing

Gene Editing



Infectious Disease Research

**www.revvity.com**



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# Contents

# Foreword

## Revitalising the NHS: A 10-Year Vision for Healthcare Transformation

With the election of the first Labour Government in fourteen years, we find ourselves at a defining moment for the future of the NHS. While the challenges remain significant – escalating patient waitlists, workforce shortages, and a health system struggling to keep pace with rising demand – there is now an opportunity to set a decisive new direction. The Government's forthcoming 10-year plan for health and care, due to be published next year, offers the chance to address these issues head-on, revitalising the NHS and ensuring it is fit for the future.

The challenges cannot be overstated. The NHS, once a symbol of world-class health services, now faces an unprecedented crisis. Staff shortages, growing waiting times, and declining patient satisfaction have created an unsustainable situation, one that threatens not only the efficacy of healthcare services but also the economic stability of the nation. Poor health outcomes and inequality in access undermine productivity and growth, compounding the urgency for reform.

The new Secretary of State for Health and Social Care, Rt Hon Wes Streeting MP has boldly highlighted on several occasions that the NHS is "broken". Fixing the system is clearly a priority for the Government and as it reviews the responses to the Change NHS: A Health Service Fit for the Future consultation, there is much to consider when it comes to one of the most important and overlooked parts of the healthcare system – diagnostics. The Chancellor's vision to implement the Government's ambitious Missions-based approach through industrial strategies is bold. If delivered through language that resonates with all levels of the NHS, the Life Sciences Industrial Strategy will be transformational for patient outcomes. Combined with a fresh approach towards diagnostics and investment, the UK has the opportunity to become world leading.

Yet, this is not a time for despair. The potential for transformation has never been greater. Innovation in technology, a renewed focus on collaboration, and a fundamental rethinking of how we deliver health services can pave the way for a brighter future. By leveraging cutting-edge diagnostic tools, data-driven decision-making, and advances in genomics, we can drive efficiency, improve outcomes, and create a more sustainable healthcare system.

Revvity, whose contributions were essential to this report, exemplifies this transformative approach. Their work in diagnostics, genomics, and data integration, combined with a collaborative ethos, showcases the potential of the life sciences and diagnostics sector to lead the way. By bridging the gap between research, technology, and patient care, they are already delivering tangible benefits for both patients and health systems.

This report serves not only as a call to action but also as a roadmap for change. It offers practical recommendations to support the Government's vision of a reformed NHS – one that gives mental and physical health equal priority, invests in prevention, and places patients at its heart.

The road ahead will not be easy, but with collective effort, sustained innovation, and a commitment to transformation, we can secure a thriving future for the NHS and the millions of people who depend on it. This report sets out ideas for a decade of progress, driven by partnership and ambition.

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# About Revvity

At Revvity, we are daring to reimagine how diseases can be identified, treated, and cured. By working collaboratively with academics, drug developers, clinicians, clinical labs and governments, we are creating the foundations for change to improve the lives of people around the world.

Revvity is a US-headquartered company that provides health science solutions, technologies, expertise, and services that deliver complete workflows from discovery to development, and diagnosis to cure. With 2023 revenue of more than $2.7 billion, and over 11,000 employees, Revvity serves customers across pharmaceutical and biotech industries, diagnostic labs, academia, and governments. It is part of the S&P 500 index and has customers in more than 190 countries.

At Revvity, our vision is to make that world a reality by empowering scientists to reimagine healthcare. We provide the technology and services that support:

- Affordable treatments that reach the market rapidly. Today, our technologies help to ensure the right drugs are selected for development, by implementing better biological models or identifying root causes of disease – reducing risk and cost.
- Screen /diagnose diseases, support maternal and newborn health and next generation sequencing for newborn screening research.
- Novel therapeutic development to help treat and cure disease. Our technologies and expertise help develop new drug modalities and bring revolutionary cell and gene therapy treatments to patients all over the world.
- Precision medicine approaches – matching patients to the best treatment for them. Today, our technologies help to identify critical biomarkers that can be used for clinical assessments so that each individual gets the right drug, at the right time.

Our life sciences and diagnostics portfolios are built on a legacy of innovation, Revvity's technology platforms – centred on cell, protein, and genomic analysis – currently support more than 45,000 peer-reviewed publications every year and provide solutions to help tackle the huge investments required to bring a drug to market. This is why we see our solutions being applied in over 90 per cent of the world's top 50 academic institutions and pharmaceutical companies.

## Revvity technologies support over 45,000 life sciences publications annually

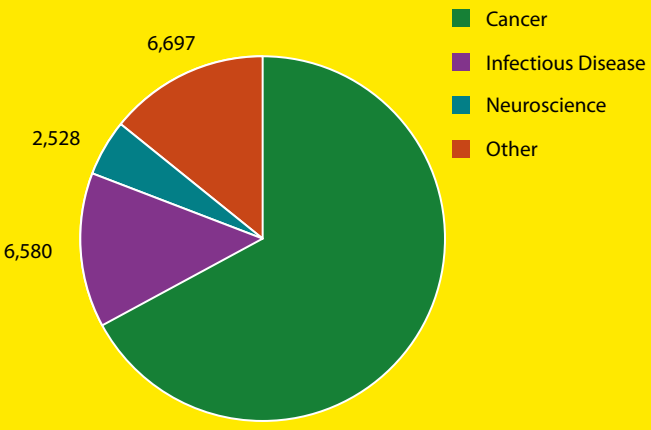



*Figure 1: Illustrates the number of peer-reviewed publications that utilize Revvity´s life sciences technologies to understand disease better.*

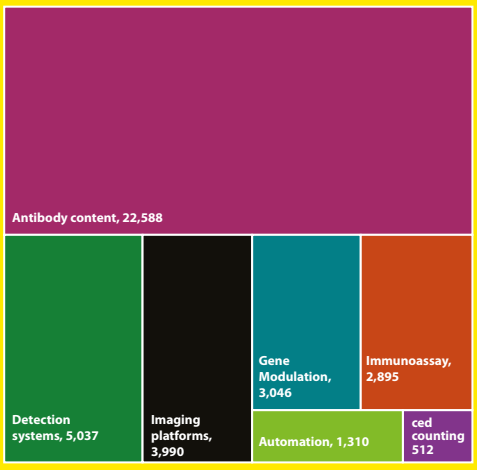



*Figure 2: Provides a breakdown of the 45,000 life sciences peer reviewed publications in 2022 by technology area.*

## Revvity in the UK

Revvity is proud to have a strong presence in the UK, with a Scientific Centre of Excellence for Genomic Insight in Cambridge, a diagnostics facility in Oxford, a manufacturing facility in Llantrisant, an immunodiagnostics site in Boldon, and a clinical grade genomics service lab in Cheshire. Together, these facilities not only contribute to the UK's economy and life sciences sector but also bring jobs and investment to five locations across the country. Revvity also provides products and services to academia, biotech, and pharmaceutical companies to support research and development in the UK, as well as diagnostic tools and laboratory services to the health service. These products and services empower scientists and clinicians and ultimately, contribute to better outcomes for patients.

Revvity recognises the UK is a hub for life sciences and is eager to see this sector thrive. For this to happen, it is crucial that life sciences remain a priority for both current and future UK governments. We welcome the commitments made by the new Government as they look to publish their Life Sciences Industrial Strategy. However, as focus shifts towards post-election implementation, Revvity would like the new Government to consider and enact the recommendations highlighted within this report.

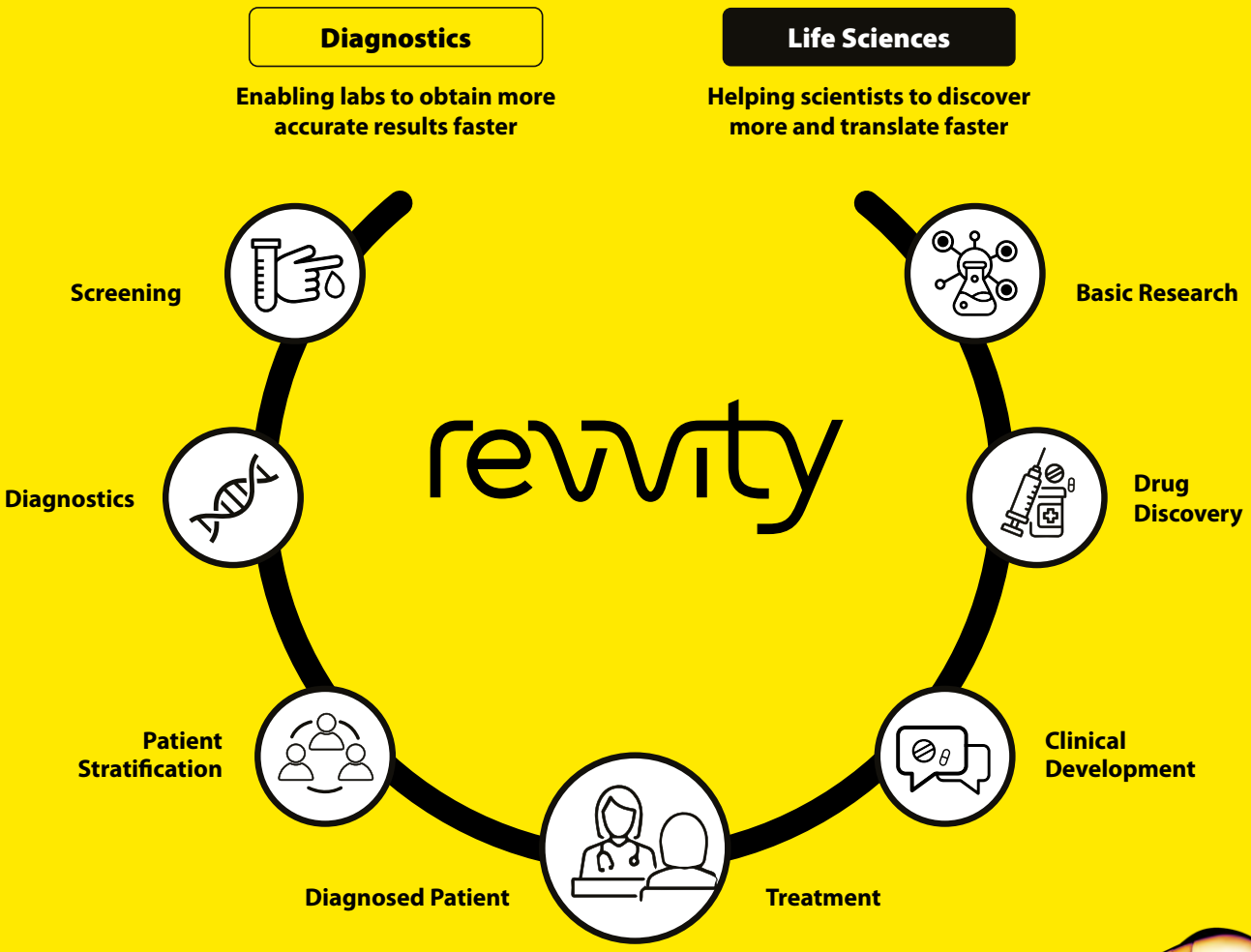



*Figure 3: Illustrates how Revvity´s life sciences and diagnostic business work together to empower scientists and clinicians to reimagine healthcare*

# Headline Recommendations

As the new Government looks to implement their plans to fix the "broken" NHS, the following headline recommendations cover critical areas such as clinical research, diagnostics, data utilisation, AI integration, and investment in the life sciences sector – aiming to drive innovation and improve patient outcomes within the NHS.

- **Ensure Rapid Access to Diagnostic Tools:** to address the backlog of patients waiting for key diagnostic tests, resource community diagnostic centres (CDCs) should adequately expand their support for rare conditions. Establish streamlined national reimbursement pathways and utilise health innovation networks to facilitate rapid evaluation and adoption of proven diagnostic technologies.

- **Implement the Lord O'Shaughnessy Review for Clinical Trials:** fully implementing the recommendations of the review will help maintain the UK's position as a global leader in clinical research. This includes providing more incentives for healthcare professionals to engage in clinical research.

- **Harness the Power of Data:** utilise the NHS Federated Data Platform to enable the safe and secure use of patient data for improved treatment and care. Invest in data infrastructure and workforce development to ensure health and care professionals can effectively take advantage of data for decision-making.

**Embrace the Potential of Artificial Intelligence (AI):** establish an expert panel under the AI Safety Institute to evaluate the ethical implications of AI in diagnosis and treatment. Develop evidence-based guidelines for responsible AI integration, prioritising fairness, privacy, transparency, and accountability.

**Invest in Jobs and Growth:** explore direct incentives for life sciences investment and prioritise the life sciences sector in free trade arrangements. Ensure regulatory alignment with major regulators like the EMA and FDA to enhance international collaboration and support innovation in the UK.

# Introduction

The NHS is facing unprecedented challenges. With over 7.5 million patients waiting for elective care,[1] over one in ten staff leaving the NHS between June 2022 and June 2023,[2] and an increasing demand for services[3], the NHS is facing a crisis of unparalleled proportions.

This will have an impact not only on the health service, patients, and their families but also on the economy, with the highest ever number of people economically inactive due to ill health.[4] High levels of ill health can hinder productivity and impact economic growth.[5]

Instead of a cost, spending on healthcare should be seen as an investment in the wellbeing and economic prosperity of the country. This will require a transformation, not only in how health spending is viewed but also with a fresh approach to how we tackle disease.

Today, the most successful part of the health ecosystem is when there is real collaboration between academic institutions, the healthcare system, life sciences companies, service providers, and governments – all with the common aim of improving patient outcomes.

This collaborative approach needs to be unpinned by a favourable ecosystem for the life sciences sector, stimulated by government support and investment. The significance of the life sciences sector in the UK cannot be overstated. As well as contributing to the economy, employment, and growth, it plays a crucial role in providing patients with access to cutting-edge treatments and therapies.

A 2023 report commissioned by the Association of the British Pharmaceutical Industry (APBI) calculated that if the 2021 *Life Sciences Vision*[6] were realised, it could generate £165 million in revenue, save the NHS £38 million, and reduce the disease burden by 40 per cent in every year.[7]

Ensuring a thriving life sciences sector in the UK is therefore pivotal to the NHS, the economy and, most crucially, patients. Failure to maintain competitiveness on the global stage may result in the UK being side-lined, consequently delaying patient access to innovative treatments.

A global healthcare revolution is underway, characterised by advanced diagnostic tools, novel approaches to drug development, and the more widespread adoption of whole genome sequencing (WGS), underpinned by digital technologies and AI-enabled data to bring treatments more rapidly to market. But for continued progress to happen, the entire pharmaceutical ecosystem needs to work together to reimagine healthcare and improve patient outcomes.

# 1. Embed Clinical Research at the Centre of the NHS

The UK has the infrastructure and experience to be a global leader in undertaking clinical research. With a universal health system, a world-leading academic sector, and highly skilled and engaged staff, the UK has a history of delivery in the field of clinical trials. The COVID-19 pandemic showed what the UK was capable of, delivering the world-leading RECOVERY[8] and PANORAMIC[9] trials, and the Vaccine Task Force.[10] As well as being beneficial to patients, clinical research activity in hospitals is associated with reduced mortality and improved overall care quality, as well as a positive impact on staff and organisation performance.[11] Reversing the decline in commercial clinical trials could yield £4.4 billion from life sciences companies and save the NHS £2.8 billion in pharmaceutical product costs.[12]

Revvity provides services for clinical trials, from patient stratification and sequencing to software offering deep insights into clinical trial data. Revvity are therefore keen to maintain the UK as a location for all stages and types of clinical research. For this to be achieved, the new Government must implement, in full, the Commercial Clinical Trials in the UK: the Lord O'Shaughnessy Review[13] into commercial clinical trials, as well as provide more incentives for healthcare professionals to get involved with clinical research, such as through financial incentives payable directly to their unit or department.

# 2. Ensure Rapid Access to and Uptake of Diagnostic Tools

Diagnosis is a critical element of the treatment and care pathway, and yet there were 1,611,800 patients waiting for a key diagnostic test at the end of February 2024.[14]

Revvity provides comprehensive diagnostic solutions for laboratories and clinics to enable reliable testing for a range of conditions, from infectious diseases to reproductive health to allergies.

There must be a renewed focus on early diagnosis to reduce the burden of ill health on both patients and the health system. To ensure patients can access necessary diagnostic tools at pace, the Government must ensure the 160 new Community Diagnostic Centres (CDCs) are resourced sufficiently to meet the ambitious targets they were set.[15] As well as achieving the aim of 17 million tests annually,[16] CDCs should be expanded to support those suspected to have a rare condition. This would reduce the need for patients with rare diseases having to make multiple visits to a healthcare setting for diagnostic tests.

A second challenge for diagnostics is that even tools proven to be safe and effective can struggle to get traction. Without streamlined national reimbursement pathways, manufacturers are often required to go to each organisation, conduct multiple pilots and repeatedly generating the same evidence. To drive rapid, national adoption of proven technologies, new diagnostic technologies should be rapidly evaluated, with clear guidelines for adoption and uptake. Health Innovation Networks[17] in partnership with Integrated Care Boards (ICBs) should be responsible for supporting the adoption of innovation locally, providing the connection with life sciences industries and ensuring that good practice spreads between geographies. Additionally, there should be a working group to review innovations across diagnostics and treatments to ensure that effective tools and treatments are being rapidly adopted across the health system.

**Case Study:** revvity

### Revvity's Lab in a Lab Service

Revvity's Lab in a Lab service provides a turnkey solution that assists laboratory scale and acquires new testing capabilities especially in support of rare disease diagnosis. Revvity's Lab in Lab service leverages the experience of our internal clinical laboratory service and manufacturing capabilities to setup the required laboratory service in an existing facility. In the midst of the SARS-COV-2 pandemic, Revvity[18] was able to rapidly setup and operate COVID testing facilities in the UK. Similarly Lab in a Lab services are being offered to organisations in India and the USA supporting testing areas such as prenatal and newborn screening testing.

# 3. Harness the Power of Data

The potential of healthcare data is well documented. It can speed up diagnosis, stratify patients and contribute to the development of personalised treatments. It is also a valuable asset for the NHS.[19]

Unfortunately, health data often exists in isolated, fragmented, and incomplete silos, reducing its potential effectiveness. The real value of data comes to light when diverse datasets are connected and made interoperable.

For Revvity, data is at the heart of everything we do. Cellular assays and imaging technologies have substantially increased in throughput capabilities in recent years. This is good news for researchers who need to generate robust data because it enables them to obtain more data in less time while maintaining high data quality. This can be converted into actionable insights for our research and clinical customers. That is why Revvity want to see a greater use of joined-up data sets across the NHS. The new Government must seize the opportunity offered by the NHS Federated Data Platform[20] to use patient data in a safe and secure way, to deliver better treatment and care.

Once the data infrastructure is in place, it is crucial that health and care staff can benefit from this resource, and thus, NHS England must ensure that alongside investment in infrastructure, there is commensurate investment in the workforce to widen data skills across the NHS for all health and care professionals.


# 4. Embrace the Potential of Artificial Intelligence (AI)

AI has the potential to transform every aspect of our lives, including healthcare. No one can fully grasp what AI may be capable of, but Revvity uses machine learning (ML) and deep learning (DL) to provide advanced data analysis capabilities with our imaging software. Revvity scientists have also designed and implemented ML/DL methods in the software to automate cellular imaging analysis tasks and optimise the segmentation and classification of image data.[21] Revvity is accelerating the pace of drug development by enabling the implementation and analysis of more predictive advanced model systems.

Looking ahead, Revvity believes AI holds the promise of enabling early prediction and detection of diseases, including neurodegenerative disorders like Alzheimer's disease. This could fundamentally change the way such conditions are diagnosed and potentially treated. However, it is crucial that AI is used responsibly in every instance, with a strong emphasis on fairness, privacy, transparency, and accountability. To ensure these principles are adhered to, an expert panel should be established under the AI Safety Institute to evaluate the ethical implications of using AI in diagnosis and treatment. Furthermore, there must be robust, evidence-based guidelines for the responsible integration of AI, reinforcing the importance of fairness, privacy, transparency, and accountability.

# 5. Invest in Jobs and Growth

Revvity welcomes the new Government's commitment to maintaining tax incentives for the life sciences sector, such as R&D tax credit and the Patent Box. Revvity has twelve sites in the UK, with five sites focused on R&D and manufacturing, and an attractive incentive landscape is vital in ensuring investors feel welcomed in the UK. However, fragmented manufacturing investment incentives in the UK, contrasted with new incentives for the manufacture of essential generic medicines in countries such as France, Germany, and the USA, as well as current disparities in batch testing recognition between the UK and EU, have diminished the attractiveness of the UK as a manufacturing location. To address this, the new Government should explore flexibilities to introduce direct incentives for life sciences investment.

They should also prioritise the life sciences sector in Free Trade Arrangements, with the aim of supporting innovation through intellectual property protection, regulatory coherence, and the removal of barriers to services, such as those that support data transfers and the movement of skilled workers.

# 6. Implement a Cutting-edge Regulatory Environment

It is vital that all diagnostics, devices, and medicines used in the UK have been validated as safe and effective. Getting products to market at pace is beneficial for patients and health systems, but also for economic growth. UK regulators should capitalise on new 'regulatory freedoms' post-Brexit to expedite the delivery of cutting-edge products to patients.

The new Government should realign its strategic approach with those of major regulators, such as the European Medicines Agency (EMA) and the US Food and Drug Administration (FDA), to enhance international collaboration. While co-operation in existing regulatory processes, including medicines and devices, is beneficial, The UK can become a global leader in innovative and rapidly developing technologies, like AI, digital therapeutics, and Advanced Therapy Medicinal Products (ATMPs). By developing an appealing regulatory framework that enables companies to launch innovative products while upholding stringent quality and safety standards, the UK can establish itself as a prime location for investment, thereby stimulating job creation and economic growth.

# 7. Driving Uptake of New Technologies

The UK is falling behind comparable countries when it comes to the uptake of new medicines and technologies.[22] This does not go unnoticed in international boardrooms, where investment decisions take into account the local ecosystem for the adoption of innovation. The UK faces particular barriers when it comes to the adoption of diagnostics, digital, and other health tech, as there is no consistent reimbursement and adoption pathway, comparable to that for medicines.

Standardised pathways for evaluating non-medicine technologies that are known to offer value for money and enhance patient choice would be welcomed, particularly by manufacturers of diagnostics and digital health technologies. Where appropriate, this pathway should adopt a tiered assessment approach based on risk, with a transparent linkage between assessment outcomes and reimbursement decisions.

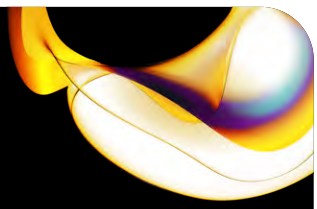
# 8. Introduce the Latest Scientific Breakthroughs to Identify and Treat Rare Diseases

Taken together, rare diseases are not that rare. There are over 10,000 genetic and rare diseases affecting up to 400 million people worldwide.[23] Eighty per cent of rare diseases have genetic origins,[24] making genomic testing a key tool in supporting healthcare professionals in finding a clinical diagnosis or recommending treatments and care that may improve patient outcomes.

However, with many rare and ultra-rare conditions affecting only a few individuals globally, it can be difficult to categorise conditions and identify individuals for clinical research. As a leading international exemplar, Revvity is embedded in the rare disease ecosystem, providing innovative, end-to-end workflows for biomarker identification and characterisation, as well as applications that enable laboratories to translate insights into decisions rapidly and efficiently. Revvity also provides support to current NHS Genomic Laboratory Hubs to help clear the current analysis backlog through innovative systems.

Whilst the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) in England collects and analyses data on rare diseases and congenital anomalies to support healthcare planning, research, and treatment, a truly comprehensive national rare disease registry is urgently needed. Such a registry would require enhanced integration and alignment across existing services to capture a broader spectrum of patient data. This could include not only genotypes, phenotypes, and patient histories of confirmed rare disease diagnoses but also other critical datasets, such as biochemical profiles, imaging results, and other clinical metrics generated for each patient. By serving as a robust database for research and a repository of treatments and outcomes, this registry could significantly advance understanding, foster innovation in treatments, and enable precise healthcare planning. Additionally, it would provide a more accurate quantification of the true burden of rare diseases and their overall impact on society, enhancing the ability to address these conditions effectively. Additionally, it would provide a more accurate quantification of the true burden of rare diseases and their overall impact on society, enhancing the ability to address these conditions effectively.

**Case Study:** revvity



## Whole Genome Sequencing to Support Rare Disease Patients

A baby became severely ill immediately after birth and sadly died at four months with no confirmed diagnosis. Analysis of his whole genome uncovered a severe metabolic disorder due to the inability to take vitamin B12 inside cells, explaining his illness. This enabled a predictive test to be offered to his younger brother within one week of his birth. The younger child was diagnosed with the same disorder but was able to be treated with weekly vitamin B12 injections to prevent the progression of the disease.

# 9. Support the Development of Cell and Gene Therapies

Some disease occurs when a segment of DNA is damaged, causing the body to malfunction. By making changes to DNA on a cellular level, it may be possible to treat the root cause of disease. These cell and gene therapies, also known as also known as ATMPs, are amongst the most complex and costly to develop, manufacture and deploy.

Life sciences companies such as Revvity supports the development of these treatments through gene editing techniques for DNA manipulation, the development of viral vector technologies, and services to optimise gene delivery, as well as providing FMP-grade reagents for therapeutic manufacture and developing novel assays to facilitate the assessment of new therapeutic modalities.

However, once they are developed, there can be issues with deployment, particularly for point-of-care manufactured products, which are those medicines produced in clinical settings and immediately delivered to patients.[25] Additionally, there are often challenges with system readiness and capacity for these treatments, as well as reimbursement delays, due to the intrinsic long-term uncertainty associated with these treatments.[26]

While cancer often dominates discussions about personalised medicine, it is critical to recognise the parallel importance of personalised approaches in rare diseases. For example, CRISPR-based therapeutic approaches in rare diseases require therapies to be tailored to the individual patient. This is because each therapy demands unique RNA guides or specific corrections to disease-causing mutations. The availability of cell and gene therapy approaches has further enabled the acceleration of new therapy development for rare diseases. Historically, new therapies for rare diseases have lagged behind due to the high cost of development and regulatory approval relative to the limited number of patients who can benefit. These advances exemplify how personalised medicine is not only transforming oncology but also unlocking new possibilities for treating rare diseases at a genetic level. Integrating this perspective into the development of advanced therapies ensures a holistic approach, addressing the diverse needs of patients and further reinforcing the need for a UK-wide advanced therapy taskforce to drive innovation and deployment.

To address this and ensure access and uptake of the latest cell and gene therapies, the new UK Government should create an advanced therapy taskforce focused on the development and deployment of advanced therapies as a strategic UK-wide health policy priority.

**Case Study:** revvity

### Revvity Licenses PinPoint™ Base Editing Platform Technology to AstraZeneca

Announced in 2023, AstraZeneca licensed the technology to develop universal donor cells, which, in turn, will be used as the foundation for various targeted therapies, such as cell therapies for the treatment of immune-mediated diseases and cancer.

Revvity's[27] new PinPoint™ base editing platform technology allows researchers to make small, precise changes in genomic DNA. These higher precision edits mean the PinPoint™ technology is safer for therapeutic use as it reduces the risk of unwanted rearrangements to the genome. In addition to an improved safety profile, the modular nature of the technology allows researchers to specifically reach their gene or genes of interest without compromising
cell health.

# 10. Deliver on the Promise of Personalised Medicines

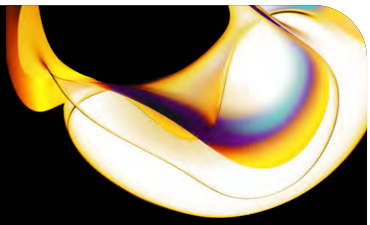
Limitations with the current one-size-fits-all approach to treatment can be seen most clearly in cancer, where, despite the achievements made in treating the disease during the past decades, resistance to classical treatments or new targeted drugs continues to be a major challenge. To combat this, researchers are trying to understand which unique genetic and environmental circumstances determine potential resistance barriers, with the goal of matching individuals to the right course of treatment or clinical trial. However, cancer biology is incredibly complex, with the average adult having over 36 trillion cells and 400 cell types,[28] each providing a different function and each cell having its own evolving agenda. Thus, unpacking the many interconnected systems requires a multidisciplined approach to build a clearer picture of patient biology to identify predictive biomarker signatures.

The complexity of cancer often means that two cancer patients with the same cancer type may not share the same set of gene mutations, meaning genetics alone may not be enough to direct therapy.[29] Recent research has shown that functional drug testing can complement molecular screening approaches to better match individuals to the right treatment for them. In short, this is achieved by taking diseased tissue from an individual and exposing it to many treatments to reveal drug sensitivities and predict an individual's clinical response.

As a leading life sciences company in the field, Revvity supports researchers by developing single-cell analysis solutions for the identification of new biomarkers and therapeutic targets, as well as end-to-end sequencing workflows and services to identify genetic markers of disease.

These techniques allow for the development of 'personalised' medicines, tailored for an individual's phenotype. However, once these 'personalised' medicines make it to market, they can be very expensive. The new Government should consider alternative models of reimbursement, such as risk and gain share partnerships or outcomes-based payments, more widely utilised for the NHS to share in both the risks and the benefits of new technologies.

Case Study:



## Revvity Drug Sensitivity Testing of 3D Ex Vivo Cultures

Researchers at the Institute for Molecular Medicine Finland (FIMM) are using automated microscopic imaging and analysis to investigate the response of patient-derived 3D cancer cell clusters to a customised panel of anti-cancer compounds. This approach aims to understand the cellular heterogeneity in cancer better and to elucidate an individual's sensitivity and resistance to different therapies. The aim is to characterise the phenotypic alterations or actions of the small molecule at the single-cell level and, from there, understand how drugs are behaving, thus gaining an understanding of the resistance of these cancer cells to certain drugs.

The Opera Phenix®30 Plus high-content screening system lets researchers quickly generate high-quality cellular responses to drug perturbations, allowing researchers to explore many drug combinations without delaying time to results. In addition, its innovative optical design and machine learning algorithms can pick out subtle differences within complex cell cultures, meaning small toxicological or potency effects can be captured early.

# 11. Encourage Widespread Use of Whole Genome Sequencing (WGS)

The 100,000 Genome Project demonstrated some of the many benefits of using whole-genome sequencing (WGS) to study and diagnose rare diseases, cancers, and infections in the UK.[31] Evidence showed WGS led to a new diagnosis in 25 per cent of the patients, and 25 per cent of these cases led to 'focused care', meaning that in addition to receiving a diagnosis, these patients received additional therapies to help manage their conditions. For certain conditions, including intellectual disability, vision, and hearing disorders, there was a higher diagnostic yield of 40–55 per cent. As a result, the NHS in England became the first national healthcare system in the world to offer WGS to people with undiagnosed rare diseases and cancer as standard care.

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.

WGS is beneficial not only in driving a diagnosis. In cancer, genomic evaluation is becoming an integral component of the diagnostic workflow, facilitating disease classification, risk stratification, and therapy selection in various cancer types.[32] Additionally, drug targets supported by human genetic evidence are twice as likely to succeed in the clinic.[33] Utilising genetic data to establish the function of a new drug target is important because, often, the biological mechanisms behind genetic variants linked to disease are unknown.

Despite the benefits, there are several 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, and therefore prevents 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 setup 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 has developed innovative, comprehensive, and customisable solutions to fast, reliable, and flexible genomic testing services that keep pace with cutting-edge technological and scientific developments, without any upfront costs. Revvity operates a global network of state-of-the-art genomic testing laboratories staffed by board-certified, experienced, clinical geneticists. Through offering genomics testing as a service, Revvity works with governments, clinical, and research institutions the opportunity to rapidly realise the benefits of this technology, with a diagnostic menu tailored to both national and local requirements. Revvity also provides an in-house model, run by the customer, enabling organisations to rapidly access the most cutting-edge genomic applications with minimal capital expenditure.[34]

Considering the many benefits, the WGS should be routinely integrated into clinical care to enable earlier identification of genetic conditions or inherited diseases, as well as for predictive and preventative healthcare.

**Case Study:** revvity

## Lowering the Barrier to Disease Diagnosis

Access to genetic testing is a big challenge, particularly in developing nations where insurance coverage is limited, forcing the public to pay out of pocket. To make genetic testing more accessible and equitable, Revvity[35] collaborates with biopharmaceutical companies focused on discovering potential treatments for rare diseases. Sponsored testing programmes, in partnership with biopharma, essentially means that the patient pays nothing or very little for genetic testing to receive a diagnosis. Through these funded collaborations, we can help lower the diagnostic barriers and provide answers for the undiagnosed.

Case Study:

## Revvity Enables the Identification of Gene Associated to Alzheimer's Progression

Advances to edit genes have revolutionised how researchers study gene function, providing previously unobtainable insights into biological pathways. More recently, gene editing is being applied to human induced pluripotent stem cells (iPSCs), which are thought to exhibit phenotypes closer to human pathology. This approach is particularly attractive to the neuroscience field, where the connection between genetics and cellular function is less clear and where therapies to treat neurodegenerative conditions, such as Alzheimer's disease, remain elusive.

In a recent study, researchers at Denali Therapeutics combined gene editing with human iPSC-derived immune cells found in the brain to explore the role of two genes that are known to be linked to the development of Alzheimer's disease. Through a series of experiments, the researchers determined the impact of two receptors on gene expression, function, and lipid metabolism. Crucially, the study revealed novel insights into the interaction of a specific protein to one of the receptors, which might be beneficial in Alzheimer's disease research and therapeutic strategies.

Dharmacon™[36] solutions allow researchers to silence gene expression within living cells robustly. When combined with our high-content imaging platforms, or antibody content from BioLegend® for flow cytometry, we are enabling researchers to identify which genes are involved in specific cellular processes.

# 12. Promote the Use of Newborn Screening

Newborn screening (NBS) plays a vital role in both saving and improving babies' lives. 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.[37]

In the UK, congenital anomalies occur in approximately 1.6% per cent of live births, with data from NCARDRS showing that around 1 in 63 live births involves a congenital anomaly.[38]

Genetic disorders and congenital anomalies affect approximately six per cent of live births in the United States and are the leading reasons for hospitalisation and mortality in infants.[39] Newborns admitted to neonatal intensive care units (NICU) with genetic disorders have longer hospitalisations and higher resource utilisation than those with other conditions. Rapid genomic testing in critically ill neonates transforms rare disease diagnosis, management, and treatment by delivering results in time to change acute medical or surgical management and inform and provide answers to families.[40] [41]

Next-generation sequencing (NGS) has the potential to revolutionise newborn screening by offering significant advantages over traditional methods. NGS allows for the simultaneous screening of multiple genetic disorders in a single test, enabling the detection of a broader range of conditions that may not be identified through standard biochemical assays. For example, Duchenne Muscular Dystrophy (DMD) can initially be screened using a CKMM assay, but the definitive diagnostic test requires sequencing to identify copy number variations and point mutations. This ability to precisely pinpoint genetic abnormalities facilitates timely intervention and treatment, which can prevent or mitigate the onset of symptoms, improve health outcomes, and reduce the overall burden of disease on patients and families.

Additionally, NGS provides high sensitivity and specificity, minimising the risk of false-positive or false-negative results and delivering more accurate and reliable outcomes. Faster turnaround times enable earlier clinical management and interventions. NGS platforms are also highly flexible and scalable,

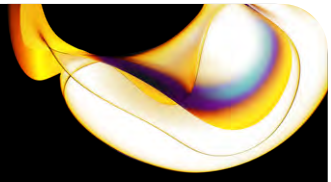
allowing healthcare providers to customise screening panels for specific populations, geographic regions, or clinical needs. This adaptability ensures that screening protocols can evolve alongside advances in our understanding of genetic diseases and emerging public health priorities, positioning NGS as a cornerstone of modern precision medicine in newborn screening.

There is already some use of WGS for newborns globally. The Generation Study in the UK is a ground-breaking research study that will sequence the genomes of 100,000 newborn babies to understand whether WGS is beneficial in diagnosing and treating genetic conditions.[42] Newborn sequencing has potential, but there are several ethical and social challenges that must be considered prior to full utilisation.

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. Revvity offers sequencing for targeted genes, multiple genes, the whole exome, or genome, and copy number variations. Revvity deliver results promptly and empower providers to use genetic testing earlier in the diagnostic process. Recent product launches include prenatal whole genome sequencing to better understand foetal anomalies and ultrarapid genomic testing for critically ill infants in the NICU who may have life-threatening disorders such as SCID, X-ALD, and Pompe disease.

Following the outcome of The Generation Study, Ministers must act on its recommendations without delay to ensure the UK is on a par with our European neighbours.  Alongside this, the NHS should carefully explore options for prenatal genetic testing under certain circumstances.

**Case Study:** revvity



## Ultra-rapid Whole Genome Sequencing

A three-day old male child presented to his physician with hypotonia, significant birth defects, and metabolic acidosis. The physician ordered Revvity's 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 *PDGA1* 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 to give this child the best possibility of an improved outcome. This chance would not have been possible without Revvity's quick turnaround time and comprehensive genetic analysis.

# Conclusion

The NHS is facing some of the most formidable challenges in its history. Yet technology presents some promising solutions. A thriving life sciences sector, supported by government investment and innovative policies, is essential for enhancing patient outcomes and driving economic growth. Revvity is at the forefront of these efforts, providing vital services and technologies that can significantly improve the quality and efficiency of healthcare delivery.

The collaborative model that includes academia, healthcare providers, life sciences companies, and government bodies has demonstrated its value and must be expanded. The recommendations put forward by Revvity emphasise the necessity for a healthcare ecosystem that embraces advanced diagnostics, innovative treatment approaches, and a robust data infrastructure to meet the demands of a global healthcare landscape undergoing rapid technological change.

By focusing on integrating these advancements within the NHS, building a supportive regulatory environment, and ensuring that healthcare professionals are equipped to utilise these technologies, the UK can be globally competitive. Moreover, by prioritising investments in life sciences, the UK will not only improve health outcomes but also stimulate economic growth, ultimately benefiting society as a whole.

# Endnotes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18 Testing services may not be licensed in accordance with the laws in all countries. The availability of specific test offerings is dependent upon laboratory location. The content of this report 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.

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27 The Pin-point™ base editing platform technology is available for clinical or diagnostic study and commercialisation under a commercial license from Revvity.

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# About Curia

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