

FUTURE
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INITIATIVE
BY ABU DHABI

The Future of Genomics

Precision Therapeutics, Value
Creation and a Discovery
Engine for Rare Diseases
and Beyond



Knowledge Contributor

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Executive Summary

Advances in genomics, AI and precision therapeutics are changing the rare disease landscape. The field is moving beyond identifying disease-causing variants towards understanding the biology they disrupt, discovering therapeutic targets and developing interventions tailored to the underlying molecular cause. Rare diseases are particularly valuable in this shift because they can reveal insights into human biology that support innovation both within and beyond the original condition. Abu Dhabi has already established important foundations for this transition, including population-scale genomics, an Emirati reference genome, genomics-enabled screening programmes, specialist clinical capabilities and pathways for access to advanced therapies.

These assets create an opportunity not only to improve outcomes for individuals within the UAE through earlier diagnosis, more accurate variant interpretation and access to precision interventions, but also to generate new biological knowledge from a population that remains underrepresented in global genomic datasets. In doing so, Abu Dhabi can both strengthen rare disease care locally and contribute globally to the understanding of genetic variation, disease mechanisms and therapeutic opportunities that remain insufficiently characterised in existing reference populations. Recent partnerships across gene editing, genomics research and rare disease care indicate an ambition to extend these capabilities into discovery, translational research and therapeutic development.

Realising this opportunity will depend on how effectively Abu Dhabi connects its genomic, clinical, research and innovation assets. Success should be measured not only by the number of genomes sequenced or partnerships established, but by whether these foundations generate better care, scientific discoveries, therapeutic opportunities and enduring local capability. With clear priorities and well-structured partnerships, Abu Dhabi could become a genomics oasis where healthcare delivery, scientific discovery and life sciences development reinforce one another.

1. Introduction: Genomics and Rare Diseases are at an Inflection Point

Overview of Rare Diseases and Genomics

More than 7,000 distinct rare diseases have been identified globally, and together they affect an estimated 300 million people worldwide, or between 3.5% and 5.9% of the global population.¹ In May 2025, the World Health Assembly recognised rare diseases as a global health priority, noting that these conditions are often complex, multisystemic, chronic and progressive, and can result in serious disability and premature death.

In the UAE, a high prevalence of inherited disorders associated with consanguinity shape the burden of rare diseases. The Catalogue for Transmission Genetics in Arabs records 665 distinct genetic conditions in Emiratis, of which 491 are classified as rare diseases, with more than half extremely rare by global standards and some unique to the Emirati population.²

Critically, much of this variation is absent from the international reference databases against which laboratories interpret genetic findings. For instance, a 2023 analysis of 1,365 gene variants reported in Emiratis found that 235 (17%) were absent from dbSNP and ClinVar as of December 2022, illustrating the historical underrepresentation of Emirati genomic variation in international reference databases.²

Closing that gap is the purpose of two complementary initiatives. The Emirati Genome Programme aims to sequence the entire Emirati population, with the Department of Health (DOH) reporting in June 2026 that more than 900,000 genomes had been sequenced.³ The Emirati Reference Genome Programme then gives clinicians a consensus blueprint of Emirati genetic diversity, allowing variants to be interpreted against a local reference rather than a foreign one.⁴

This creates both a healthcare imperative and a scientific opportunity. Underrepresentation in global reference data makes interpretation harder for Emirati individuals, but it is also where new variants, gene-disease relationships and biological mechanisms remain to be found, improving rare disease care and generating new biological knowledge locally and globally.

¹ World Health Organization, Rare diseases: a global health priority for equity and inclusion, Resolution WHA78.11, Seventy-eighth World Health Assembly, 27 May 2025

² Sami Bizzari et al., "Spectrum of genetic disorders and gene variants in the United Arab Emirates national population: insights from the CTGA database", *Frontiers in Genetics*, vol. 14, 3 May 2023, <https://doi.org/10.3389/fgene.2023.1177204>.

³ Emirates News Agency (WAM), "Department of Health – Abu Dhabi, Novartis sign strategic MoU to advance genomics research, healthcare innovation", 26 June 2026, <https://www.wam.ae/en/article/c0x4f93-department-health-abu-dhabi-novartis-sign>

⁴ Emirati Genome Program, "Emirati Genome"

"Emirati genetic variation has long been missing from the references the world uses to interpret genomes. Every genome sequenced through the Emirati Genome Program helps close that gap, giving clinicians more accurate answers for their patients and giving global science a clearer view of human diversity and disease," explains David Kavanagh, General Manager, Omics, M42.

Key Developments in the Genomics and Rare Diseases Landscape

Genomics and rare diseases innovation has historically been constrained by lengthy and costly diagnostic journeys, limited understanding of rare disease biology, small and dispersed individual populations and high barriers to therapeutic development. These barriers are beginning to shift as multiple technologies and capabilities converge:⁵

- Falling genetic sequencing costs with advanced technologies and the expansion of population-scale genomics
- Greater availability of linked genomic, phenotypic and clinical data
- Improved computing capacity and AI-enabled interpretation
- Advances in multi-omics, functional genomics, spatial biology and disease modelling
- The emergence of RNA and cell-and-gene-editing therapeutic platforms

From Variant to Value: The Genomics and Rare Diseases Discovery Chain

Together, these advances in sequencing, data generation, AI-enabled analysis, functional genomics and therapeutic platforms are shifting the field from identifying rare-disease-causing variants towards understanding rare disease biology and developing targeted interventions. While genomics remains the necessary foundation, discovery increasingly depends on complementary capabilities, which link genetic variation to biological mechanism and, ultimately, to intervention.⁶

This progression can be described as four connected shifts, each building on the one before, and each to be explored in the upcoming sections:

- From identifying disease to understanding biology
- From understanding biology to biological discovery, knowledge and innovation
- From target discovery to precision therapeutics
- From therapeutic innovation to a connected discovery ecosystem and value creation⁷

⁵ Elizabeth J. Wilk et al., "AI in variant analysis: fast track to genetic diagnoses", Human Genetics, vol. 145, article 54, 26 June 2026, doi: 10.1007/s00439-026-02847-0.

⁶ Breige McBride, "The Future of Genomics", Fios Genomics

⁷ James H. Buchanan, Ilias Goranitis and Deirdre Weymann, "The Health Economics of Genomic Technologies: A Growing Evidence Base on Value", Applied Health Economics and Health Policy, vol. 23, no. 3, 2025, pp. 331–335, doi: 10.1007/s40258-025-00970-z.

Exhibit 1: From Variant to Value – The Rare Disease Discovery Chain

Stage	Key Question	Enabling Technologies and Capabilities
 Sequencing	What genetic variants exist?	Whole Genome Sequencing, Long-Read Sequencing
 Interpretation	What do these variants mean?	AI-driven Variant Interpretation, Bioinformatics, Population Reference Genomes
 Biology	How do these variants cause disease?	Multi-omics (e.g. transcriptomics, proteomics, metabolomics), Functional Genomics, Spatial Biology, etc.
 Discovery	What pathways and targets can be identified?	Disease Modelling, CRISPR Screening, AI-enabled Target Discovery
 Therapeutics	How can we intervene?	Gene Therapy, RNA Therapeutics, Gene Editing, Cell Therapies
 Value Creation	How do we generate impact?	Learning Health Systems, Clinical Trials, Real-World Evidence, Discovery Ecosystems

2. Deep Dive into the Shifts Defining the Future of Genomics and Rare Diseases

2.1 From Identifying Disease to Understanding Biology

Early Focus Originally Limited to Variant Identification

In the early genomics era, capability was largely confined to one step of the discovery chain: identifying the genetic cause of disease. Sequencing could establish which variant an individual carried, but the tools to explain what that variant did, how it disrupted biology, or where intervention might be possible were far less developed. Identification therefore became the endpoint rather than the starting point.

Today, identifying disease-causing variants is no longer the principal limiting factor. Sequencing costs have fallen, and whole genome sequencing now resolves a substantially higher share of previously undiagnosed cases than targeted panels do.⁸ The constraint has moved downstream, from generating the genetic finding to explaining it.



⁸ The 100,000 Genomes Project Pilot Investigators, "100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care: Preliminary Report", The New England Journal of Medicine, vol. 385, no. 20, 11 November 2021, pp. 1868–1880, doi: 10.1056/NEJMoa2035790

Advanced Genomic Sequencing and Complementary Technologies Enabling Biological Insight

Answering questions about what a variant does, how it disrupts biology and where depends on genomics continuing to advance alongside a complementary set of capabilities and technologies. Genomics is not a fixed capability: long-read and whole-genome sequencing resolve variants that earlier methods could not detect, population-scale programmes build the reference base against which those variants are interpreted, and AI-driven interpretation is steadily extending what a genome can be made to explain⁹. Nevertheless, explaining what a variant does, how it disrupts biology and where intervention may be possible depends on a complementary set of technologies that enable capabilities to connect genetic variation with molecular pathways, cellular behaviour, individual phenotype and clinical outcomes.^{10,11,12} The focus therefore moves from identifying a mutation to building a richer understanding of disease biology.

Overview of Key Capabilities and Technologies

- Advanced genomic sequencing (whole genome sequencing, long-read sequencing): Detects variants earlier methods missed, and provides the reference base against which findings are interpreted
- Multi-omics (transcriptomics, proteomics, metabolomics): Shows what a variant changes at the level of gene expression, protein and metabolic function
- Spatial biology (single-cell technologies): Reveals which cell types and tissues are affected and how disease is distributed within an organ
- Functional genomics (high-throughput technologies): Tests the biological effects of genetic variants, helping identify which changes are truly driving disease
- Data and AI platforms (bioinformatics, AI, Digital twins): Integrate genomic, molecular and clinical data to identify disease-associated variants, prioritise therapeutic targets and uncover patterns at a scale beyond manual analysis

Taken together, these capabilities move the question from which variant is present to what that variant does, connecting a genetic change to the pathways it disrupts, the cells and tissues in which it acts, and the clinical picture it produces.

⁹ Chen, R., Duffy, Á. & Do, R. "Genomics of drug target prioritization for complex diseases", *Nature Reviews Genetics*, vol. 27, 2026, pp. 231–245

¹⁰ Sebastian Lunke et al., "Integrated multi-omics for rapid rare disease diagnosis on a national scale", *Nature Medicine*, vol. 29, no. 7, 2023, pp. 1681–1691, doi: 10.1038/s41591-023-02401-9

¹¹ Caroline Seydel, "Beyond cell atlases: spatial biology reveals mechanisms behind disease", *Nature Biotechnology*, vol. 43, 2025, pp. 841–844.

¹² Elizabeth J. Wilk et al., "AI in variant analysis: fast track to genetic diagnoses", *Human Genetics*, vol. 145, no. 1, article 54, 26 June 2026, doi: 10.1007/s00439-026-02847-0

"A genome without context is a question without an answer. It is sequenced once but interpreted for a lifetime. Across cancer, rare disease, and reproductive genetics, our hardest cases are rarely limited by sequencing. They are limited by context: a variant in the right gene that no global database can yet classify. Connecting genomic data to clinical history, family history, and real-world outcomes turns an uncertain finding into a definitive diagnosis, and a diagnosis into actionable evidence," observes Burjeel Holdings.

From Mutation to Mechanism to Disease Behaviour

Understanding how a variant disrupts biology is only part of the picture. The same genetic mutation can affect individuals very differently in terms of severity, age of onset, organ involvement and response to treatment. In some cases, a person carries a variant known to cause disease but never develops the condition at all. In others, everyone affected carries the same variant, yet one person is mildly affected while another is severely so, sometimes within the same family. Cystic fibrosis illustrates the point: a single-gene disorder in which more than 80 modifier genes have been shown to influence disease progression, severity and therapeutic response.^{13,14} The explanation lies beyond the causative variant itself, in modifier genes, polygenic background, regulatory variants, epigenetics and environmental exposure.

"Two people can carry the same genetic variant and experience very different health trajectories due in part to their lifestyle and environmental exposures. The clinical question is what that finding means for the individual today. At IHLAD, we look at inherited factors, alongside life course, current biology and functional status. Following these dimensions over time helps clinicians understand what may require action today, what needs monitoring and where more evidence is needed before a change is made. That is how genetics becomes more useful in caring for a person," explains Dr. Nicole Sirotin, CEO, Institute for Healthier Living Abu Dhabi (IHLAD).

A full understanding of disease therefore requires knowing not only what a variant does, but how its effects unfold in individuals over time. This depends on longitudinal clinical data, advanced phenotyping and molecular profiling rather than a single snapshot at diagnosis.¹⁵ Studying whole populations provides a truer picture, and variants once believed to always cause disease are increasingly being reclassified. For a population underrepresented in global reference data, that work has to be done locally; otherwise, it does not happen at all. As Rashed Al Qubaisi, Group Chief Operating Officer, PureHealth explains *"Connecting genomic, clinical and outcomes data can help advance genomic medicine beyond diagnosis towards earlier intervention and more personalised, lifelong care."*

¹³ Strand Life Sciences, "One Gene, Many Stories: Understanding Phenotypic Diversity in Rare Disorders"

¹⁴ Ward A, Mauleon R, Ooi CY, Rosic N. Impact of Gene Modifiers on Cystic Fibrosis Phenotypic Profiles: A Systematic Review. Hum Mutat. 2024 Oct 16;2024:6165547. doi: 10.1155/2024/6165547. PMID: 40225935; PMCID: PMC11919198.

¹⁵ Kingdom R, Wright CF, "Incomplete Penetrance and Variable Expressivity: From Clinical Studies to Population Cohorts", Frontiers in Genetics, vol. 13, 2022, doi: 10.3389/fgene.2022.920390.

Key Message

Beyond identifying which genetic variants cause disease, the next frontier will be about understanding how those variants disrupt biology and how their effects unfold differently across individuals over time.¹⁶ Genomics provides the foundation, but understanding rare diseases requires combining genomics with complementary technologies and longitudinal clinical data that explain how variants disrupt biology and how their effects unfold across individuals over time.



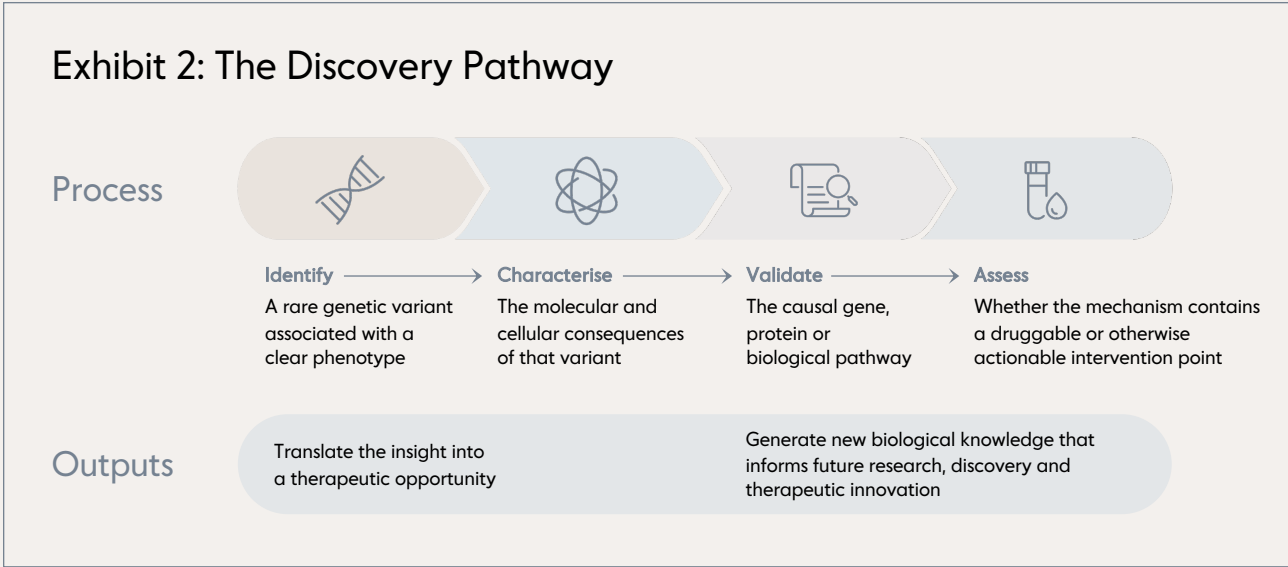
¹⁶ James Gao, "Genomics: Revolutionizing Medicine and Shaping the Future of Healthcare", International Journal of Collaborative Research on Internal Medicine and Public Health, vol. 16, no. 6, pp. 001–002, 2024, doi: 10.35248/2332-2594.24.16.06.001-002

2.2 From Understanding Biology to Discovery, Knowledge, and Innovation

From Biological Insight to Discovery Opportunities

Understanding rare disease biology produces two distinct outputs. The first is therapeutic opportunity: a validated target against which a treatment can be designed. The second is an expanding base of genomic and biological knowledge, richer variant interpretation, better-characterised pathways and a stronger reference base, all of which enable discoveries that are yet to be made.

Rare genetic conditions can function as naturally-occurring experiments in human biology. A mutation may increase, reduce or eliminate the activity of a gene or pathway, and the consequences are observable in individuals rather than inferred from a model. Studying these extreme biological states with advanced technologies can clarify gene function, reveal causal pathways, and identify potential points of intervention.¹⁷ Establishing what a gene does and how the resulting protein behaves also reveals whether that gene or protein can realistically be acted on by a medicine. The set of such targets is known as the *druggable genome*.¹⁸ The value therefore extends beyond the individual therapeutic opportunity: each discovery contributes to a growing body of biological knowledge that improves understanding of human disease, informs future research and creates new opportunities for target discovery, biomarker development, and therapeutic innovation across multiple disease areas.



¹⁷ Anne R. Pariser and William A. Gahl, "Important Role of Translational Science in Rare Disease Innovation, Discovery, and Drug Development", *Journal of General Internal Medicine*, vol. 29 (Supplement 3), 17 July 2014, pp. S804–S807, doi: 10.1007/s11606-014-2881-2

¹⁸ Oprea, T.I., Bologa, C.G., Brunak, S. et al., "Unexplored therapeutic opportunities in the human genome", *Nature Reviews Drug Discovery*, vol. 17, 2018, pp. 317–332, doi: 10.1038/nrd.2018.14.

"Each diagnosis is never about one patient. It identifies a family, and every family is an opportunity for prevention through carrier screening, preconception planning, newborn screening, and cascade testing. A single gene studied in a handful of families can reveal a mechanism relevant to millions and has repeatedly led to treatments for common conditions. The infrastructure, clinical expertise, and bioinformatics built to solve rare diseases are the foundations required for population-wide hereditary cancer screening and precision prescribing. Rare disease is where population health begins," states Burjeel Holdings.

Knowledge as a Compounding Asset

The final step is easily overlooked, but it is where much of the long-term value sits. Each characterised condition improves the interpretation of future variants, strengthens understanding of the pathways involved, and generates hypotheses that extend well beyond the original patient population. Because genes operate within shared biological pathways rather than in isolation, a target shown to be druggable in one rare condition often points to intervention opportunities in others affected by the same pathway. Therapeutic opportunities are the visible output; the accumulating body of biological knowledge, including a growing map of which targets are actionable, is the asset that compounds over time and makes subsequent discoveries faster and more likely.

Illustrative Example: Menkes Disease and the Biology of Neurodegeneration

Menkes disease is a rare, severe childhood disorder caused by mutations in the ATP7A gene, which disrupts the transport of copper throughout the body and brain. Although the disease affects only a small number of individuals, decades of research into Menkes disease transformed scientific understanding of copper transport and metal homeostasis, revealing how trace metals are regulated at the cellular level and how their dysregulation can impair neurological function.

These discoveries extended well beyond Menkes disease itself. The ATP7A pathway has since been linked to mechanisms involved in common neurodegenerative disorders, including Parkinson's disease, where abnormalities in copper metabolism and metal homeostasis are increasingly recognised as contributors to disease progression. In this way, a rare genetic disorder provided fundamental insights into biological processes that underpin much more prevalent conditions, illustrating how rare disease research can generate knowledge with broad relevance across medicine.¹⁹

¹⁹ Lee CE, Singleton KS, Wallin M, Faundez V., "Rare Genetic Diseases: Nature's Experiments on Human Development", iScience, vol. 23, no. 8, 2020, article 101419, doi: 10.1016/j.isci.2020.101419

Illustrative Example: PCSK9 and Cholesterol-lowering Therapies

Rare genetic variants in the PCSK9 gene provided critical insights into the biological regulation of cholesterol. Researchers observed that individuals carrying loss-of-function variants in PCSK9 had markedly lower levels of low-density lipoprotein cholesterol (LDL-C) and a substantially reduced risk of cardiovascular disease. These naturally occurring genetic experiments validated PCSK9 as a key regulator of cholesterol metabolism and identified it as an attractive therapeutic target. The discovery subsequently led to the development of PCSK9 inhibitors, a new class of cholesterol-lowering medicines now used globally to reduce cardiovascular risk.²⁰

Key Message

Rare diseases offer a window into human biology, transforming genomic insights into a growing pipeline of discoveries, therapeutic opportunities and innovations that will shape the future of medicine.



²⁰ Grejtakova, D., Boronova, I., Bernasovska, J. Bellosta, S. "PCSK9 and Lipid Metabolism: Genetic Variants, Current Therapies, and Cardiovascular Outcomes", Cardiovascular Drugs and Therapy, doi: 10.1007/s10557-024-07599-5

2.3 From Target Discovery to Precision Therapeutics

A validated target doesn't directly translate to a treatment or intervention. Rare disease biology yields causal mechanisms and identifies which of them are realistically druggable. What follows is the harder question of what a health system does with that knowledge, i.e. how a mechanism becomes an intervention, how the right individual is matched to it, and how quickly that has to happen to be worth doing. This is where genomics shifts to becoming a basis for decisions.

The Traditional Model: Managing Symptoms

For many rare diseases, treatment has traditionally focused on slowing disease progression, reducing complications, managing clinical manifestations, and preserving quality of life. Where the causal mechanism is unknown, supportive care is the only available and effective response to reduce the burden of disease. While these approaches remain important, they often do not alter the biological process responsible for disease. Symptomatic care manages the consequences of a molecular fault without altering the fault itself, so it must be sustained for as long as the person lives with the condition.

The Emerging Model: Precision Intervention

As disease mechanisms and therapeutic targets become clearer, treatments can increasingly be designed around a specific gene, pathway, cell type, or molecular defect. The shift is from treating the consequences of disease toward intervening in its underlying biology. Where symptomatic care manages the effects of a faulty gene indefinitely, a precision intervention aims to correct, replace, silence or bypass the fault itself, in some cases through a single administration rather than through lifelong therapy.²¹ This shift is being enabled by a new generation of therapeutic platforms, including gene therapy, RNA therapeutics, cell therapies, and gene editing, each designed to intervene at different points between a genetic alteration and its clinical consequence.

Three properties distinguish this model, and each has consequences for how a health system must be organized around it:

- **Durability:** The clinical effect is intended to persist well beyond the period of administration
- **Irreversibility:** A one-time intervention cannot be titrated, paused or withdrawn if the indication proves wrong. The margin for error in patient selection is therefore narrower than in conventional prescribing
- **Selection:** Once a therapy exists, the difficulty is ensuring the right individual can be identified, confirmed and treated

²¹ Sommariva, Elena, Milena Bellin and Chiara Di Resta, "Advance in Genomics of Rare Genetic Diseases", *Biomolecules*, vol. 13, no. 10, 2023, article 1441, doi: 10.3390/biom13101441.

The Role of Risk Stratification in Precision Medicine

Precision medicine is often described as delivering the right treatment to the right patient. In practice, however, this objective can only be achieved when genomic findings are translated into clear clinical actions. Risk stratification provides this mechanism by converting complex genomic information into practical pathways for care.²²

Every individual's genome contains a wide range of findings, including pharmacogenomic variants, carrier status for inherited conditions, variants of uncertain significance, and, in some cases, medically actionable findings unrelated to the original reason for testing. Not all findings require the same degree of intervention. Some can be managed through routine prescribing adjustments, while others may warrant specialist genetics consultation, genetic counselling, additional diagnostic investigations, or advanced therapeutic interventions.

Risk stratification combines genomic, clinical, and phenotypic information, increasingly supported by AI-enabled analytics and predictive models, to guide treatment decisions across three areas:²³

- Treatment eligibility: Genetic variants, molecular targets, and biomarkers can determine whether a patient is likely to benefit from a therapy
- Risk of adverse outcomes: Genetic factors can predict toxicity, serious adverse reactions or reduced treatment effectiveness, enabling treatment plans to be adjusted proactively
- Treatment timing and prioritisation: Risk stratification helps identify which patients require immediate intervention, ongoing monitoring, or early preventive treatment

Applied consistently, risk stratification enables precision therapeutics to move beyond individual cases and into routine clinical practice. It is only meaningful if each tier resolves to a defined action. By making these decisions explicit and standardized, risk stratification supports consistent clinical practice, reduces unwarranted variation between providers, and helps healthcare systems scale precision medicine safely and efficiently.

²² Wang, R.C.; Wang, Z. "Precision Medicine: Disease Subtyping and Tailored Treatment". *Cancers* 2023, 15, 3837. <https://doi.org/10.3390/cancers15153837>

²³ Sakhaa Alsaedi, Michihiro Ogasawara, Mohammed Alarawi, Xin Gao, Takashi Gojobori, "AI-powered precision medicine: utilizing genetic risk factor optimization to revolutionize healthcare", *NAR Genomics and Bioinformatics*, vol. 7, no. 2, 2025, article lqaf038, doi: 10.1093/nargab/lqaf038

Key Equity and Ethics Considerations

Access, Readiness and Sustainability

Risk stratification is usually presented as a clinical method. In rare diseases and precision therapeutics, it is also a mechanism for determining who is identified, prioritised and receives access to specialised interventions. That gives it an ethical dimension that must be examined rather than assumed. Risk criteria can determine who receives intervention, and should therefore be explicit, published, clinically justified and subject to regular review. As treatment recommendations increasingly depend on risk stratification, equitable access becomes a system-wide requirement rather than an individual clinical consideration. Healthcare systems will need to ensure equitable access to coordinated referral pathways, centres of excellence, a workforce equipped to interpret and communicate genomic results, clinical research infrastructure and related support services.

Ethics, Trust and Responsible Use

Precision medicine depends on the collection, analysis and sharing of highly sensitive genetic information, and participation is voluntary. Realising the benefits of precision medicine therefore requires strong safeguards around privacy, consent, data governance and the responsible use of genomic information.

Genomic findings can have implications not only for individual patients but also for their relatives, raising questions about disclosure, data ownership and informed consent. As technologies such as gene editing become more sophisticated, healthcare systems will also need clear ethical frameworks to guide their appropriate use²⁴. Maintaining public trust will be as important to the future of precision medicine as scientific and technological progress.

Key Message

The future of precision medicine lies not only in developing new therapeutics, but in building health systems capable of identifying the right individuals, matching them to the right interventions and acting early enough for those interventions to have maximum impact.

²⁴ Maria Siermann, et.al, "Emerging and evolving values in the changing landscape of genomics", *Frontiers in Genetics*, 2025, doi: 10.3389/fgene.2025.1566291.

2.4 From Therapeutic Innovation to a Connected Ecosystem and Value Creation

The Limits of Isolated Capability

Each one of the aforementioned shifts describes a capability: sequencing at scale, interpreting variants, understanding biology, validating targets, developing precision therapies. None of them delivers value in isolation. A genomic sequencing programme that does not connect to clinical care generates data rather than diagnoses. A research institution without access to longitudinal outcomes can characterise a variant but not observe how it behaves over time. A therapeutic developer without access to genetically characterised patients has a target, but no route to a trial. The limiting factor is not the technology or the infrastructure — it's the connection, governance, and the willingness to share methods, tools, and interpretation across institutional boundaries.

The next phase of genomics and rare disease innovation therefore depends not only on new technologies and a growing knowledge base, but on the ability to connect healthcare delivery, research, genomic data, and therapeutic development into a continuous cycle of discovery and improvement.²⁵



²⁵ Manolio TA, Rider R, Bult CJ, et al. "Advancing the science of genomic learning healthcare systems", *Frontiers in Genetics*, vol. 15, 2024, article 1484915, doi: 10.3389/fgene.2024.1484915.

The Stakeholders that Make Up a Discovery Ecosystem

Discovery ecosystems are built from key organisations and stakeholders because no one institution holds all the ingredients: patients, data, scientific capability, therapeutic development capacity, and capital.

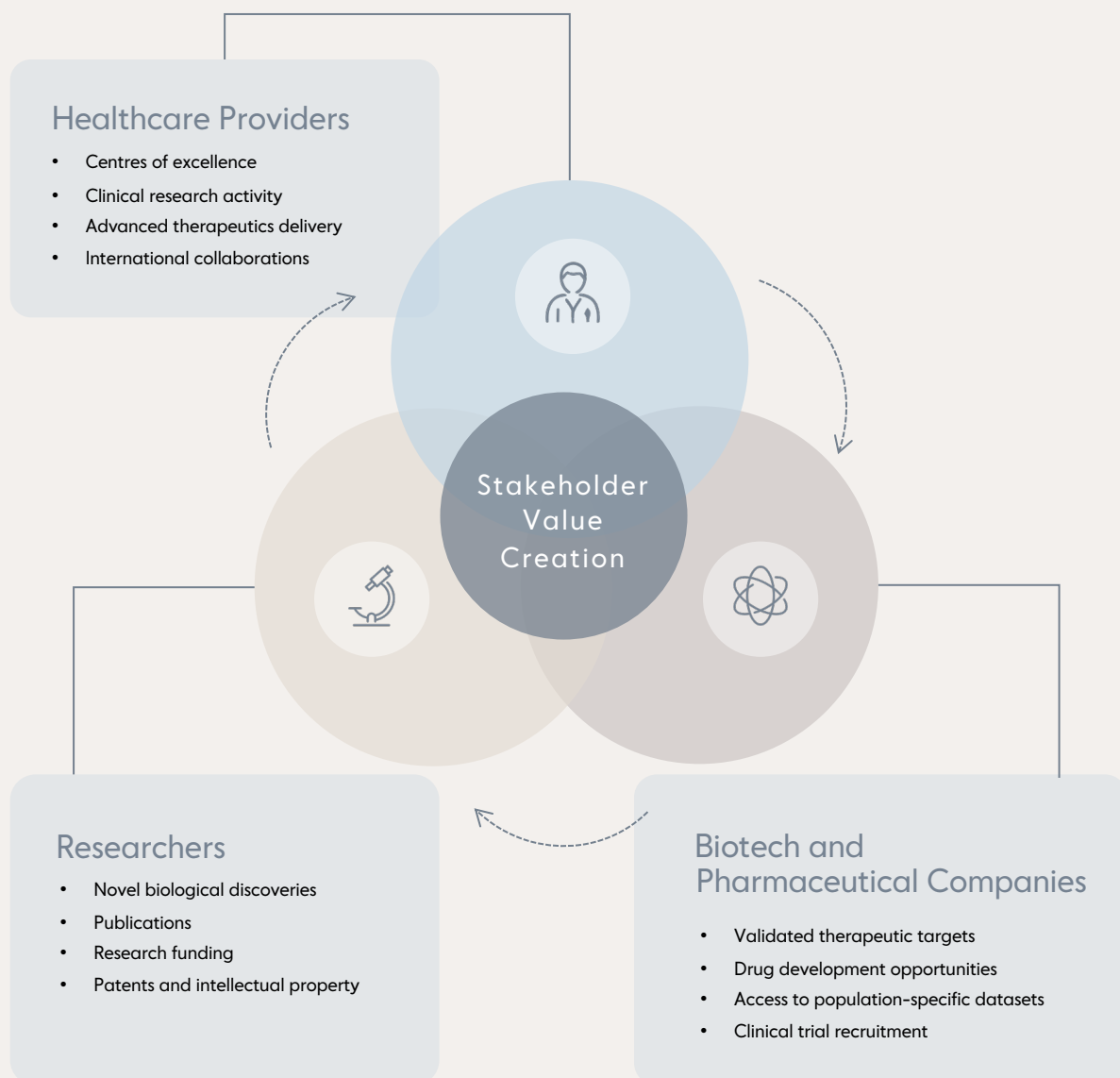
	Patients and families	Provide the clinical presentation, consent, and longitudinal participation on which everything else depends
	Healthcare providers and specialist rare disease networks	Diagnose, treat, and generate the clinical data that links genotype to outcome
	Population genomics programmes, registries, and biobanks	Supply variant data, biological samples, and the reference base for interpretation
	Research institutions and translational laboratories	Characterise mechanism, validate targets, and convert observation into evidence
	AI, data, and bioinformatics platforms	Interpret variants at scale and connect genomic, molecular and clinical datasets
	Biotechnology, pharmaceutical, and advanced-therapeutics developers	Translate validated targets into therapies and manufacture them
	Regulators, payers, investors, and economic-development entities	Set the conditions under which discovery becomes an approved, funded, and accessible therapy

The challenge is not assembling these stakeholders, but connecting them in a way that allows knowledge, data, and innovation to flow between them.

Value Creation Across the Ecosystem

Where genomics, clinical care, research, and therapeutic development are connected, rare diseases become more than a cost of care: they generate scientific, clinical and economic value through intellectual property, novel therapeutic targets, clinical trial activity, biotechnology innovation, research partnerships, and investment attraction. This value is shared across the ecosystem, with each stakeholder gaining something different.

Exhibit 3: Stakeholder Value Creation



Key Orchestration Requirements

Realising this value requires a set of enabling capabilities that allow stakeholders, data, and resources to operate as a connected ecosystem:

1. Interoperable genomic, molecular, and outcomes data so that information generated in one setting is usable in another
2. Clear governance for consent, data, discovery, and value
3. Shared research priorities and defined routes from clinical questions to translational programmes
4. Infrastructure for validation, trials, manufacturing, and real-world evidence generation
5. Partnership and incentive models that align health, scientific, and economic outcomes



Success Story: Genomics England | Building a Connected Genomics Ecosystem

Genomics England was established by the UK Department of Health in 2013 to deliver the 100,000 Genomes Project on behalf of the NHS, sequencing 100,000 genomes from approximately 85,000 patients with rare disease or cancer. While the programme is often recognised for its sequencing scale, its more significant achievement was institutional: it assembled the orchestration layer that allowed patients, clinicians, laboratories, researchers, and industry to operate around shared infrastructure, common standards, and a clear governance model.

Interoperable genomic, molecular, and outcomes data:

Interoperability in the programme was a design decision taken at the point of collection, standardising how clinical and genomic data were captured, rather than a technical fix applied later. Thirteen NHS Genomic Medicine Centres, recruited patients and recorded clinical information against a single model for testing, data capture, and reporting, ensuring that a record created in one centre carried the same structure and meaning as one created in another.²⁶ Sequencing was consolidated within one dedicated centre operated by Illumina and analysed through a common automated pipeline, so genomic outputs were returned in identical formats for every participant irrespective of where they entered the system. Genomic data, phenotypic data and longitudinal medical histories, including linked hospital, cancer treatment, and mental health records, were then held against shared data models and a common participant identifier, allowing them to be joined reliably across sources.

Clear governance:

Participation is consent-based, and de-identified data is held in the National Genomic Research Library (NGRL), a secure national repository of genomic and health data built by Genomics England with NHS England.²⁷ Researchers analyse this data only inside the Research Environment, Genomics England's secure cloud-based analysis platform, no individual patient-level data can leave it, and any export of analytical results passes through a controlled review process before release.²⁸ Alongside this, clinical governance sits with NHS England, where a Genomics Clinical Reference Group advises on policy and oversees annual review of the national genomic test directory, and a People and Communities Forum of patient and public representatives meets quarterly to shape services.²⁹

Shared research priorities:

The programme set explicit national objectives from the outset: benefit current patients, establish a genomic medicine service inside the NHS, enable new scientific discovery, and stimulate a domestic genomics industry.³⁰ Those objectives have since been translated into standing priority areas. Under the 2022 national genomics strategy, NHS England established eight Genomic Networks of Excellence in defined priority areas including rare and inherited disease, prenatal genomic medicine, pharmacogenomics, and genomics AI³¹. Each is a partnership between the NHS, academia, the third sector and industry, tasked with generating the evidence and adoption model needed to inform commissioning decisions giving clinical questions a defined route into translational programmes and, from there, into routine service.

²⁶ Genomics England, "100,000 Genomes Project", Genomics England. Available at: 100,000 Genomes Project | Genomics England (accessed 23 September 2026).

²⁷ Genomics England, "How your data is used", Genomics England. Available at: How your data is used | Genomics England

²⁸ Genomics England, "The Research Environment", Genomics England. Available at: The Research Environment | Genomics England

²⁹ NHS England, "Governance", NHS England. Available at: Governance | NHS England

³⁰ NHS England, "NHS Genomic Medicine Service", NHS England

³¹ NHS England, "NHS Genomic Networks of Excellence", NHS England.

Infrastructure:

The programme was carried by a national network of NHS Genomic Medicine Centres; a dedicated sequencing centre operated by Illumina; an automated analytics platform returning standardised whole genome analyses to the NHS; and the National Genomic Research Library, accessed through the secure Research Environment. This infrastructure was retained and became the foundation for the NHS Genomic Medicine Service, which made whole genome sequencing part of routine care.

Partnership and incentive models:

Delivery depended on aligning organisations with different mandates - Genomics England, established by the Department of Health in 2013; NHS England and its trusts; a commercial sequencing partner; and more than 3,000 researchers, alongside 1,500 NHS staff and 85,000 participants. Academic, biotechnology and pharmaceutical researchers can all apply to analyse the linked data under the same governed conditions, giving industry a genuine route to discovery while patient-level data stays protected³². This is the mechanism by which one of the programme's founding aims, building a domestic genomics industry, is pursued without trading away public trust.

Key Lesson

The UK experience demonstrates that the greatest value of a national genomics programme lies not in the number of genomes sequenced, but in the ability to orchestrate a connected ecosystem that links healthcare, data, research, and innovation, turning genomic knowledge into a continuous cycle of discovery, therapeutic development, and improved patient outcomes.

3. Abu Dhabi's Opportunity: From Genomics Capability to a Genomics Oasis

The burden of inherited and rare genetic conditions in Abu Dhabi creates both a healthcare imperative and a scientific opportunity. Meeting the imperative requires earlier diagnosis, specialist expertise, and access to advanced therapies. The opportunity goes further. Because the Emirati population remains underrepresented in global genomic reference data, studying disease in Abu Dhabi can reveal variants, mechanisms, and therapeutic targets that are not visible elsewhere. As population-scale genomic and clinical data accumulate, they can improve care for patients in the emirate while closing gaps in global rare disease knowledge.

Realising this opportunity would propel Abu Dhabi in becoming a *Genomics Oasis*: a place where population-specific data, clinical expertise, research capability, therapeutic development, and investment are concentrated and connected closely enough that a finding in one part of the system can be acted on in another.

The five orchestration requirements introduced in Section 2.4 provide a practical framework for assessing Abu Dhabi's readiness to make this transition, and what the emirate should prioritise next.

³² NHS England, "NHS Genomic Medicine Service Research Collaborative", NHS England.

3.1 Abu Dhabi's Readiness for a Connected Discovery Ecosystem

As discussed in Section 2.4, the long-term value of genomics is not created by individual capabilities in isolation, but by the ability to connect those capabilities into a coordinated ecosystem that continuously generates clinical, scientific, and economic value. Viewed through the five orchestration requirements outlined earlier, Abu Dhabi has made significant progress in building the foundations of such an ecosystem, while several areas remain important priorities for the next phase of development.

Orchestration Requirement 1 – Interoperable Genomic, Molecular and Outcomes Data

Abu Dhabi has already built many of the data foundations that an interoperable genomics ecosystem needs. Genomic data are generated at population scale through the Emirati Genome Programme.³³ Samples are transferred to the Omics Centre of Excellence for sequencing and analysis, which concentrates genomic data generation in a specialised facility.³⁴ The data are then interpreted against a population-specific reference through the Emirati Reference Genome Platform. Clinical data are exchanged across public and private providers through Malaffi, the emirate's health information exchange, which connects all hospitals into unified patient records.³⁵ Genomic insight is already reaching the point of care: clinicians can access pharmacogenomic reports for consenting Emirati Genome Programme participants through Malaffi.³⁶

Abu Dhabi has also created the environments needed to turn these data into research. The DOH Trusted Research Environment grants approved researchers governed access to linked genotypic, phenotypic, and clinical data for around 660,000 participants.³⁷ Access is overseen by Data Access Committees, and the environment supports cross-institution research through federated study models. The Abu Dhabi Biobank adds an additional layer by linking biological samples with longitudinal clinical, genomic, and lifestyle data.³⁸ Together, these assets give Abu Dhabi the core ingredients that the Genomics England example showed to be decisive: linked data, a secure research environment, and governed access.

The task ahead is to extend this strength as the ecosystem grows. As more partners, programmes, and data sources join the ecosystem, the common data standards and linkage methods that connect genomic and clinical records should be clearly documented and openly shared with them. That allows new data sources, including partner clinical trials, the Biobank, and the expanding rare diseases registry, to adopt the same standards from the outset rather than being aligned later.

³³ Emirati Genome Programme, "Emirati Genome Program", Emirati Genome Programme.

³⁴ G42, "G42 Healthcare inaugurates Omics Centre of Excellence", G42

³⁵ SNOMED International, "SNOMED International Recognizes Abu Dhabi Health Data Services for Leadership in Clinical Data Standardization", SNOMED International.

³⁶ Malaffi, "About Malaffi", Abu Dhabi Health Data Services

³⁷ Department of Health – Abu Dhabi, "Accelerating Drug Discovery and Precision Health in Abu Dhabi", DoH Trusted Research Environment.

³⁸ Abu Dhabi Media Office, "Department of Health – Abu Dhabi and M42 inaugurate Abu Dhabi Biobank", Abu Dhabi Media Office, 3 April 2026.

For rare diseases, each patient's clinical features should also be recorded consistently and in sufficient detail across providers, ideally using a shared vocabulary for describing symptoms and signs. Rare conditions are often recognised through patterns shared by only a few patients, so consistent, detailed recording makes those patterns easier to find and link to their genetic cause.

"Sequencing the population at scale was the first step. The real value of the Emirati Genome Program lies in what it makes possible next: linking genomes with clinical records and biological samples in a way that is consistent, secure, and open to trusted partners. That is what turns a population-scale dataset into a platform for discovery, and ultimately for therapies developed with and for the people," notes David Kavanagh, General Manager, Omics, M42.

Orchestration Requirement 2 – Clear Governance for Data, Discovery, and Value

Abu Dhabi has established a comprehensive and layered governance framework for genomics. At the federal level, Federal Decree-Law No. 49 of 2023 regulates the collection, analysis and storage of genomic data and requires informed consent prior to genetic testing, supporting the ethical and responsible use of genomic data³⁹. Within Abu Dhabi, the DOH has published dedicated policies on genomics, precision medicine, biobanking and its Trusted Research Environment, alongside guidelines for clinical and translational research in genomics.

As Abu Dhabi moves further into research and development, advanced therapy manufacturing and AI-enabled discovery, increasingly alongside international partners, its governance framework for this will need to continue evolving, particularly in the context of discoveries that move from research towards commercialisation. How should AI models and digital twins trained on genomic and health data be validated before they inform clinical or development decisions? What quality and safety standards should apply to locally-manufactured gene and cell therapies? How should gene-editing research be overseen? Under what conditions can data, or models derived from them, be shared with international partners? Answering these questions early, in step with leading international regulators, would give partners the certainty they need to invest in Abu Dhabi while protecting patients and individuals and maintaining public trust.

Orchestration Requirement 3 – Shared Research Priorities

Abu Dhabi has already set out the direction for its research ecosystem. DOH lists cancer and rare diseases among its priority disease areas, and precision medicine, advanced therapies, drug discovery and genetics and epigenetics among its priority research areas. Recent partnerships have begun to turn this direction into focused programmes. The Rare Diseases Centre of Excellence, activated with AstraZeneca and PureHealth, focuses on rare metabolic, nephrological and haematologic disorders.

³⁹ United Arab Emirates. Federal Decree-Law No. 49 of 2023 Regulating the Use of the Human Genome. UAE Legislation. Issued 2 October 2023; effective 1 December 2023.

"Through our collaboration with the Department of Health – Abu Dhabi and AstraZeneca on the Rare Diseases Centre of Excellence, we are bringing together global expertise and PureHealth's integrated healthcare capabilities to advance more personalised care. This includes SEHA's role in supporting patients throughout their journey, from genomic findings and diagnosis to treatment and lifelong follow-up," explains Rashed Al Qubaisi, Group Chief Operating Officer of PureHealth.

The collaboration with Mammoth Biosciences is reported to centre on a gene-editing candidate for rare diseases, alongside local manufacturing and workforce development.⁴⁰

As the number of partnerships grows, the next step is to connect these individual programmes into a coherent rare disease research portfolio. "Rare diseases" is a broad category. The Catalogue for Transmission Genetics in Arabs alone records 491 rare conditions in Emiratis, so choices will need to be made about where to focus first. Abu Dhabi's own data assets offer a strong evidence base for these choices. Population-scale genomic data and linked clinical records can show which pathogenic variants are most frequent in the population, which conditions carry the greatest burden and where diagnosis or treatment gaps are largest. Combining this evidence with scientific opportunity, feasibility and potential patient benefit would give a transparent basis for prioritisation, while also helping ensure that new partnerships reinforce one another rather than develop in parallel.

Orchestration Requirement 4 – Infrastructure for Validation, Trials, and Real-world Evidence

Abu Dhabi already has several components of the infrastructure needed to move discoveries towards patients. These include specialist rare disease care pathways, rapid regulatory access to advanced therapies, the Rare Diseases Centre of Excellence with its commitment to real-world evidence generation and partnerships that extend into gene-editing research and advanced therapy manufacturing. Because these capabilities have largely been developed through separate programmes and agreements, the opportunity now is to connect them into a single route that takes a promising finding through functional validation, clinical investigation, regulatory review, therapeutic delivery and long-term outcome measurement.

The priority is therefore to connect existing infrastructure rather than simply add to it. For each prioritized rare disease condition, the stages from discovery to intervention should be defined in advance. That means naming the bioinformatics and computational platforms, biobanking resources, validation laboratories, trial sites, manufacturing arrangements and real-world evidence systems it will rely on, with clear decision points for whether it progresses. Mapping infrastructure against specific programmes also shows where genuine capacity and expertise gaps exist, so future investment goes where it's needed rather than being spread across disconnected assets. Defining the route this way also shows where AI can shorten it. Clinical trials are one of the hardest stages for rare diseases because so few patients are available to take part as study subjects.

⁴⁰ M42, "Abu Dhabi Advances Gene Editing Therapies Targeting Rare Diseases Through Strategic Global Collaboration", M42, 25 June 2026.

Abu Dhabi's linked genomic and longitudinal health data provide a strong foundation for digital twins: virtual models of patients or disease, built from real data, that can simulate how patients might respond to a therapy. Their most established use is in clinical trials, where they can help researchers reach reliable findings with fewer patients, which is valuable when every patient enrolled matters. Broader uses, such as testing therapies virtually before trials begin, are promising but still need further validation and regulatory acceptance.

Orchestration Requirement 5 – Partnership and Incentive Models

Partnerships are central to Abu Dhabi's genomics ambitions, because no single institution has all the capabilities a connected ecosystem needs. At the BIO International Convention 2026 in San Diego, DOH took part in several partnerships and agreements across genomics, AI, advanced therapeutics, biotechnology, and precision medicine. These partnerships reflect a wider ambition to take part in more of the research, development and manufacturing value chain, not only to supply data and patients.

The Health, Endurance, Longevity and Medicine (HELM) cluster, launched in 2025 by the Department of Economic Development, the Abu Dhabi Investment Office, and the DOH, was set up to support this shift as an integrated platform for the research, development, manufacturing and commercialisation of biotechnology and new medicines. Above all, partnerships should be aligned with Abu Dhabi's agreed priorities for genomics and rare diseases.

Linking each partnership to the priority-setting process described earlier would focus effort where Abu Dhabi can have the greatest impact.

It would also turn broad frameworks into programmes with a named condition, cohort, site, and timeline. Beyond this alignment, key elements determine how much value partnerships leave behind:

- Governance that enables action: Each major partnership needs a clear owner, a clear decision process and a way to resolve issues
- Connected assets: Partnership research should run through Abu Dhabi's existing shared infrastructure, such as the DOH Trusted Research Environment, and follow the Abu Dhabi's common data and interoperability standards
- Terms that align outcomes: Data access, intellectual property, commercialisation, and patient access should be agreed at the outset. Incentives can link what partners gain to what they contribute

- **Capability and value built together:** The strongest partnerships co-develop therapies with local institutions, rather than only running trials or accessing data in Abu Dhabi. Building in training, technology transfer, and shared ownership of intellectual property and returns lets both sides benefit from what they create, with Abu Dhabi as a long-term development partner rather than simply a host

The real measure of a partnership is what remains once it ends: patients treated, knowledge generated, and expertise, intellectual property, and manufacturing capability anchored in Abu Dhabi.

3.2 From Foundations to a Discovery Ecosystem: Priorities for the Next Phase

Abu Dhabi enters the next phase from a position of strength. It has built the foundations that a discovery ecosystem depends on. The challenge ahead is therefore less about building new capability and more about directing what already exists. Key priorities will shape this shift:

- **Set a shared rare disease portfolio to focus the ecosystem:** Abu Dhabi's data assets are already being used to find disease-causing variants and improve variant interpretation for the Emirati population. The next step is to agree a shared portfolio of priority rare diseases, selected on population need, scientific opportunity, and potential patient benefit. The portfolio would then guide decisions across the ecosystem: which research is funded, where infrastructure is built, what expertise is required, which care pathways come first, and which partnerships are pursued. That way, resources go where they will have the greatest impact. Choosing priorities on the basis of need would also mean that conditions with less commercial appeal are not left behind.
- **Keep governance evolving with the ecosystem:** As research, advanced therapy manufacturing and AI-enabled discovery expand, governance will need to keep pace without slowing the activity it oversees. The aim is to make the right thing easy to do safely. That means clear and predictable processes for data access and approvals, with review in proportion to risk, so that low-risk research moves quickly and the strongest scrutiny is reserved for the most sensitive uses. It also means regular review of policies and standards, so that new technologies such as AI tools and novel gene therapies are covered as they emerge rather than after the fact. Clarity on how intellectual property applies to discoveries derived from Abu Dhabi's genomic data is part of this. Partners are more likely to invest in R&D when they know from the outset how discoveries will be owned and shared. Throughout, consent, privacy and transparency remain the basis of public trust, and that trust is what allows the ecosystem to grow.

- **Connect infrastructure into one route from discovery to patient:** Abu Dhabi already has many of the steps needed to turn a discovery into a treatment, from validation and clinical trials to manufacturing, regulatory review, and specialist care. The opportunity is to connect these steps for each priority condition, so that a promising finding can move through them without delays between stages. In practice, this means setting out in advance where a finding will be validated, where it will be trialled, how a therapy would be manufactured and approved, how patients would access it, and how their outcomes would be tracked over time. It also means making sure each stage has the capabilities it needs: the right talent, technologies, facilities, and data systems. Clear decision points along the way would show whether a programme should progress, change course or stop, so that resources stay focused on the most promising discoveries.

"Workforce capability must evolve in parallel. This requires expanded access to genetic services, multidisciplinary collaboration and ongoing upskilling of clinicians across both primary and specialist care," notes Mediclinic Middle East.

- **Structure partnerships to build value locally:** Every major partnership should be tied to the agreed priorities and set up under a standard framework. That framework would turn existing governance policies into consistent terms for data access, intellectual property, publication and benefit-sharing, and would include incentives and commitments that build local capability across R&D and manufacturing.
- **Close the loop between discovery and care:** Abu Dhabi already has the channels for a continuous cycle, including the Emirati Reference Genome Platform, Malaffi, the rare diseases registry and the Trusted Research Environment. Connecting them deliberately would let new findings update variant classifications, clinicians' reports, and screening programmes. Outcomes from treated patients would then flow back into research and shape the next set of priorities.

Together, these priorities move Abu Dhabi from building genomic capability to directing it, with a shared rare disease portfolio setting the direction and the other priorities turning it into discoveries, therapies, and lasting value.



3.3 Tracking Progress and Measuring Impact

Progress towards a *Genomics Oasis* should be judged by outcomes, not activity. Counting genomes sequenced, partnerships signed or projects launched shows that the ecosystem is busy. It doesn't show that it is working. Measurement should therefore operate at two levels. The first level is the ecosystem itself, assessed against the five orchestration requirements:

Data Can genomic, clinical and outcomes data be linked and reused across organisations?	• Share of rare disease patients with linked genomic, clinical, and longitudinal outcomes data • Number of providers and data sources, including partner trials and the Biobank, contributing to common data standards • Consistency and completeness of clinical feature (phenotype) recording for rare disease patients across providers
Governance Is it keeping pace with new activity while maintaining public trust?	• Time from research or data access application to decision • Frequency with which policies and standards are reviewed and updated for new technologies • Public and participant confidence in how genomic data are used, and participation and consent rates in genomic programmes
Priorities Are research and partnerships aligned with the rare disease portfolio?	• Share of research funding and partnership activity directed to portfolio conditions • Number of portfolio conditions with an active research programme, cohort, and timeline • Inclusion of high-need conditions with less commercial appeal
Infrastructure Are priority conditions moving along the route from discovery to patient?	• Number of portfolio conditions progressing through each stage, from validation to trial, approval, and delivery • Time taken to move between stages. • Clinical trials and advanced therapies manufactured locally for rare diseases • Patients identified, referred, and treated through rare disease pathways
Partnerships Are they leaving lasting capability in Abu Dhabi?	• Share of partnerships tied to portfolio conditions and meeting agreed milestones • Local researchers, clinicians, and manufacturing staff trained or leading work through partnerships • Intellectual property, companies, and manufacturing capability generated in Abu Dhabi

These are early indicators. They show whether the conditions for discovery are in place before results appear. The second level is impact, measured across the health, scientific, and economic outcomes set out in Section 3.4. Health impact should remain the primary measure: above all, a shorter time to diagnosis and earlier access to effective treatment for people living with rare conditions.

Baselines and targets for these measures would need data and input from DOH and ecosystem partners. Reviewing progress regularly, and reassessing Abu Dhabi against the five requirements as the ecosystem grows, would show where priorities need to shift.

3.4 Realising the Vision of a *Genomics Oasis*

By acting on the priorities set out above, with the DOH orchestrating the ecosystem, Abu Dhabi has the opportunity to evolve into a true *Genomics Oasis*: an ecosystem in which care, research, and innovation continuously strengthen one another. Its significance would extend well beyond the emirate. Because Emirati genetic variation remains underrepresented in global datasets, discoveries made in Abu Dhabi could improve rare disease care locally while filling gaps in global genomic knowledge. This value would be realised across three dimensions:

Health Outcomes	
• Earlier and more accurate diagnosis of rare diseases • Reduced diagnostic odyssey for patients and families • More personalised prevention, treatment, and long-term disease management	• Greater access to advanced therapeutics, including gene and RNA-based therapies • Improved quality of life and health outcomes for patients • Reduced healthcare utilisation and avoidable costs associated with delayed diagnosis
Scientific Outcomes	
• Discovery of novel disease-causing variants, genes, and biological pathways not observable in populations already represented in global datasets • Population-specific insights that improve understanding of disease in the UAE and broader regional populations	• Reclassification of variants whose significance cannot be resolved against foreign reference data • Increased participation in international research collaborations and multicentre studies • Accelerated translation of discoveries into diagnostics, therapeutics, and clinical practice • Strengthened leadership in genomics and rare disease research.
Economic Outcomes	
• Growth of biotechnology and precision medicine companies • Expansion of clinical trial and translational research activity • Generation of intellectual property and innovations	• Attraction of international investment, partnerships, and talent • Development of a highly skilled life sciences workforce • Creation of new high-value jobs across research, clinical care, data science and biotechnology manufacturing

Over time, the clearest sign of a *Genomics Oasis* will be what Abu Dhabi exports: variant classifications that laboratories elsewhere rely on, disease mechanisms first described in Emirati families, and therapies developed and manufactured in the emirate for patients far beyond it.

4. Conclusion

Rare diseases can serve as a catalyst for a connected genomics discovery ecosystem, improving patient outcomes while generating biological knowledge and new therapeutic opportunities. For Abu Dhabi, the opportunity is to build on its population genomics, clinical capabilities and emerging partnerships to move beyond generating genomic data towards translating it into discovery and innovation.

Realising this ambition will depend on connecting the emirate's genomic, clinical, research and therapeutic capabilities through clear priorities, trusted governance and well-structured partnerships. If these elements work together, Abu Dhabi can become a genomics oasis where better care, scientific discovery and life sciences development reinforce one another, while positioning the emirate as a globally recognised contributor to genomics innovation and rare disease research.

More importantly, Abu Dhabi has the opportunity to contribute something of global significance. The Emirati population remains underrepresented in international genomic reference datasets, creating opportunities to identify genetic variation, disease mechanisms and biological insights that are not yet well captured elsewhere. Through population-scale genomics, linked clinical data and partnerships spanning research, therapeutics and care delivery, Abu Dhabi can help close important gaps in the global understanding of rare diseases while accelerating the development of new diagnostics, therapies and intervention pathways.

"Genomics is changing what is possible in healthcare, enabling us to move from identifying genetic variation to understanding disease biology and developing more precise interventions. Abu Dhabi has established strong foundations through population-scale genomics, an Emirati reference genome, screening programmes, and specialist care pathways. Our next step is to connect these capabilities more closely with research, therapeutic development, and clinical care, so that this knowledge leads to earlier diagnosis, more personalised prevention and treatment, and better outcomes for patients. This is particularly important for rare diseases, where each new insight can improve diagnosis, inform more personalised treatment, and expand our understanding of human biology. By contributing knowledge from the Emirati population, which remains underrepresented in global genomic datasets, Abu Dhabi can strengthen care locally, help close important gaps in global research, and accelerate the discovery of new diagnostics and therapies for patients worldwide,"

said Her Excellency Dr Noura Khamis Al Ghaithi,
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