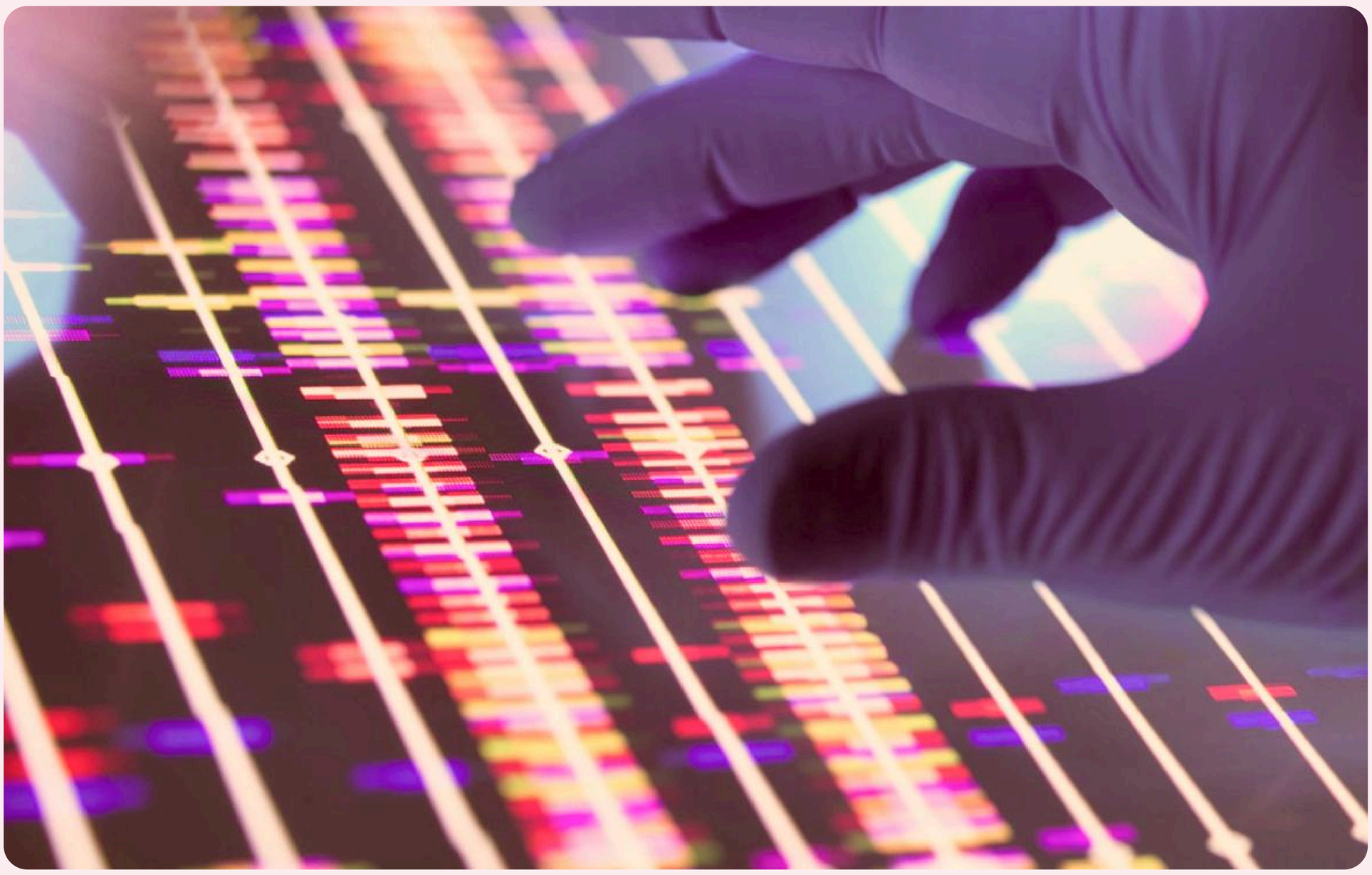
Sano®

REPORT

Diagnostics, data, and site readiness:

What world-leading CGT programs get right



Introduction

Cell and gene therapy (CGT) programs frequently reach an inflection point where scientific feasibility is established and clinical execution becomes the primary determinant of success. In practice, this period may be characterized by structural constraints in how patients are identified, how evidence is generated and sustained, and how clinical sites are equipped to deliver increasingly complex therapies.

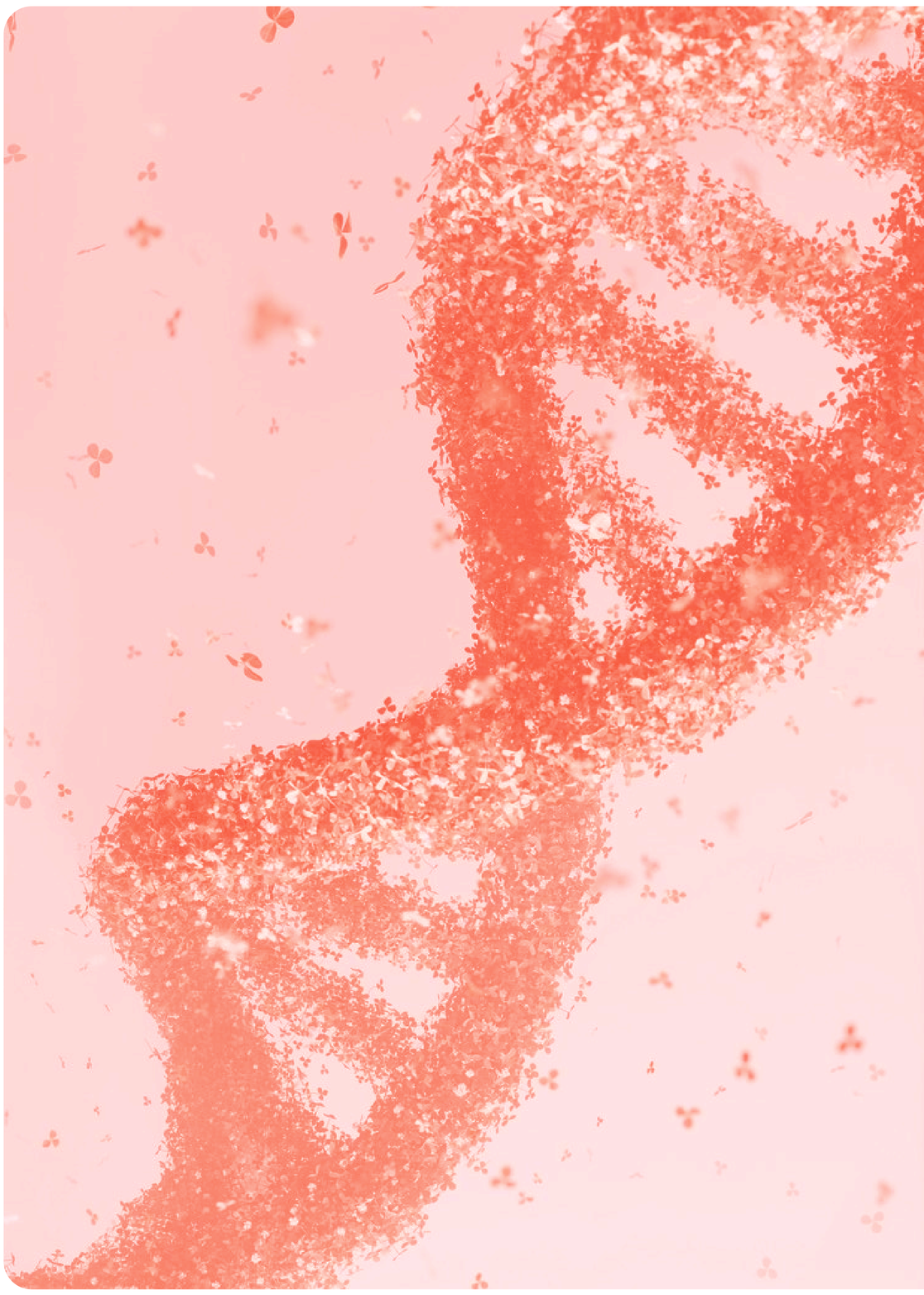
Rare disease programs may begin with an incomplete and fragmented view of the treatable patient population. Molecular confirmation remains slow and inconsistent in real-world care pathways, limiting early visibility into eligible patients. Patients with rare diseases typically experience a diagnostic delay of up to 6 years.[1] For sponsors, these delays compress recruitment timelines, reduce the number of patients identified in time for intervention, and increase heterogeneity in baseline disease stage at enrollment.

At the same time, evidence expectations for CGT extend well beyond trial readout. The FDA recommends a long-term follow-up period of 15 years to monitor for delayed adverse events following gene therapy administration.[2] This creates significant operational pressure as sponsors must establish a durable data infrastructure, sustain patient engagement, and ensure continuity across sites and care settings.

Clinical trial execution itself is becoming more constrained. A study by the Tufts Center for the Study of Drug Development showed that 70% of site staff reported that trials have become much more difficult to manage.[3] Furthermore, a WCG survey revealed that 50% of sites reported reducing the number of studies they agreed to participate in during the previous year.[4] These pressures are intensified in CGT programs, where highly specialized procedures and long-term follow-up obligations exceed the capabilities of many sites.

When diagnostics, data, and site readiness are planned and executed as separate operational streams, delays begin to accumulate. Patient identification occurs too late to support timely enrollment. Eligibility criteria must be amended as real-world diagnostic availability becomes apparent. Data required for interpretation or regulatory context must be assembled retrospectively across systems that were not designed to connect. Sites face operational demands that exceed their capabilities once protocols are active. These issues extend timelines, increase cost, and reduce the predictability of CGT development.

Taken together, these realities mean that CGT programs are more likely to be successful if sponsors treat diagnostics, longitudinal data, and site capability as crucial infrastructure from the start.



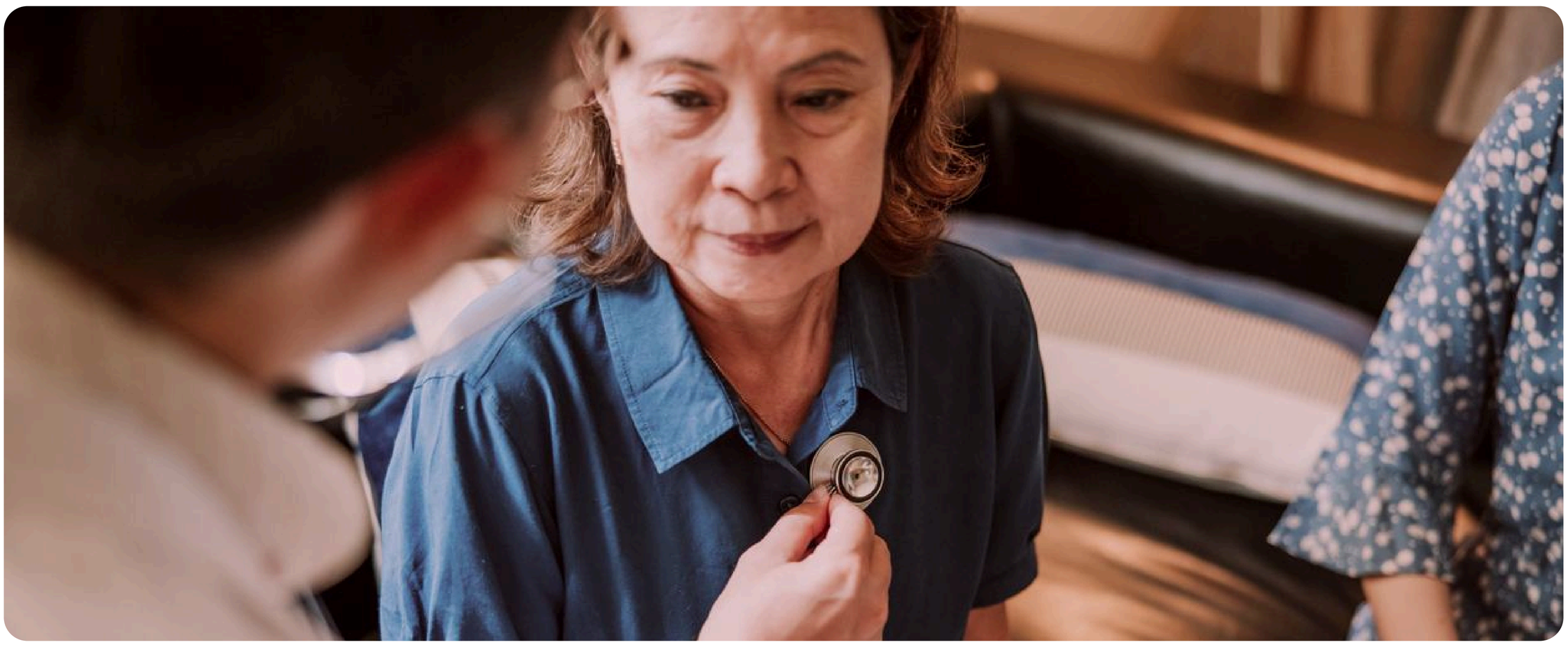
Diagnostics done well

Diagnosis is the first step in a patient journey that may culminate in participation in a CGT trial. If diagnostic pathways are disjointed and treated as distinct from referral pathways and enrollment, sponsors are likely to encounter major difficulties as handoffs slow confirmation and reduce visibility into the eligible population.

Patient identification challenges are further compounded by uneven use of genetic testing across healthcare settings. Testing decisions are influenced by provider familiarity with rare diseases and access to technology. This affects eligibility confirmation, increases screen failures, and shifts enrollment timelines. In recognition of this, various successful CGT programs have centered around diagnostics.

For instance, Sarepta Therapeutics has developed several gene therapies for Duchenne muscular dystrophy (DMD). In parallel, they sponsor Decode Duchenne, a genetic testing program that provides free genetic testing for individuals in the Duchenne and Becker muscular dystrophy community in the US and Canada.[5] The program can reduce access friction to molecular confirmation, which is a prerequisite for eligibility and for matching patients to trial opportunities.

For programs like Novartis' Zolgensma for spinal muscular atrophy (SMA) in which the patient population is newborns, the most efficient approach is to take advantage of systematic newborn screening. This also maximizes treatment efficacy as presymptomatic infants who were treated with Zolgensma avoided permanent damage.[6]



Data foundations done well

Fragmentation across data sources, study phases, and care settings limits the ability to use patient data longitudinally, particularly when early studies, trial outcomes, and long-term follow-up are not designed to connect.

In rare disease, there are often critical knowledge gaps that may make it difficult to design effective trials. Accordingly, there have been intensive efforts to establish natural history datasets that can clarify disease progression. This data can be used to optimize endpoint selection and other trial parameters. Patient data can also be used as external comparators in small trials.

An example is Généthon's DMD gene therapy program (GNT0004), which used a parallel disease natural history study to contextualize functional trajectory and support interpretation of treated outcomes.[7] In 2025, the company reported positive functional outcomes in treated patients compared with decline in untreated patients in the parallel natural history study.[8] This is an example of natural history data functioning as structured context for rare disease CGT development and for clinically meaningful endpoint interpretation.

How Sano Genetics supports integrated CGT infrastructure

Sano Genetics works with CGT programs to support coordinated patient identification, longitudinal data generation, and site engagement. Rather than addressing diagnostics, data, and site readiness as separate activities, Sano provides infrastructure that connects these elements across the development lifecycle. This integrated approach is designed to reduce fragmentation between identification, evidence generation, and delivery, and to support CGT programs from early planning through long-term follow-up.

Site readiness done well

CGT protocols place substantial operational demands on clinical sites, including complex product handling, specialized procedures, and long-term patient monitoring. These requirements affect both trial feasibility and patient safety and cannot be met through site activation alone.

In many programs, site capability is addressed late in development and in isolation from diagnostic strategy and data planning. This fragmentation leads to misalignment between protocol requirements and real-world site operations. Activation is delayed as sites work to close unforeseen gaps, eligibility workflows break down when diagnostic access varies, and data quality suffers when sites are not equipped to support longitudinal follow-up. These issues restrict enrollment and introduce operational variability that is difficult to correct once trials are underway.

Programs that deliver CGT successfully assess site capability early and in coordination with diagnostics and data strategy. Site selection and preparation account for procedural expertise, infrastructure, and the ability to sustain follow-up over time. By treating site readiness as an integrated component of program design rather than a downstream operational step, these programs reduce execution risk and support more consistent delivery.

Conclusion

Diagnostics, data generation, and site readiness function as a shared operational infrastructure in CGT development. When these elements are designed in isolation, programs face delays in patient identification, challenges in evidence interpretation, and constraints in trial execution that are difficult to resolve once studies are underway.

The examples in this report show that leading CGT programs account for these dependencies early. Diagnostic pathways are aligned with enrollment strategy. Data foundations are designed to support longitudinal use across development phases. Site capabilities are assessed in relation to protocol demands before activation. These design choices reduce downstream operational risk and support more predictable execution.

For sponsors, this has implications for when decisions are made. Choices about diagnostics, data architecture, and site readiness must be addressed before protocols are finalized and recruitment begins. Once enrollment stalls or sites struggle to execute, opportunities to fix core weaknesses are limited. Treating these elements as integrated infrastructure at the outset supports feasibility, interpretability, and long-term delivery of CGT programs.

To find out how Sano can help you simplify your precision medicine research, get in touch.

Get in touch

References

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