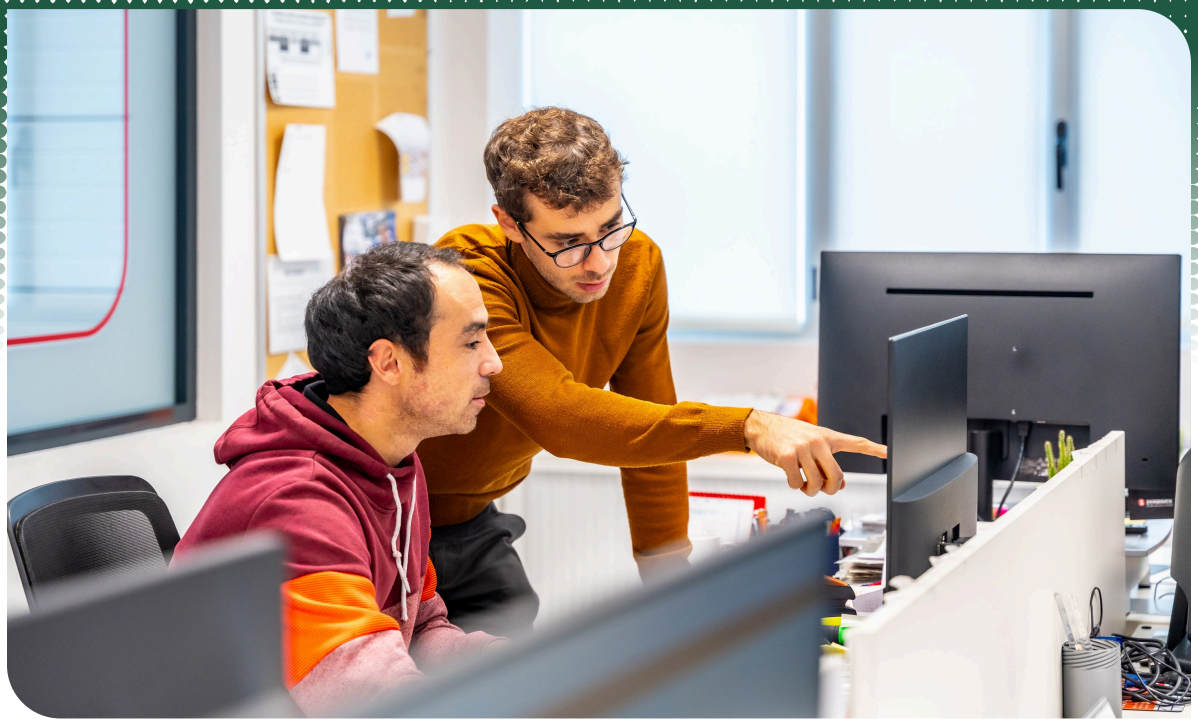
Case Study

# Transforming Rare Disease Treatment Decisions Through Curated Genomic Intelligence

## A Collaboration Between Rancho BioSciences and Rady Children's Institute for Genomic Medicine (RCIGM)



Every year, up to 10% of the nearly 3.6 million newborns in the United States are hospitalized, and many require intensive care. About one third of these cases have an underlying genetic condition that may respond to targeted treatment. In these critical moments, clinicians need rapid access to trusted, evidence-based insights to guide treatment decisions.

RCIGM developed the Gene to Treatment platform to deliver fast, accurate clinical guidance for rare genetic diseases. Rancho BioSciences partnered with RCIGM to build the scientific data foundation that powers this capability through comprehensive knowledge mining, evidence curation, and rigorous quality control. The result is a high-fidelity treatment knowledge base that supports real-time clinical decision making and sets a new standard for evidence readiness in genomic medicine.

# The Challenge

Treatment evidence for rare diseases is fragmented across thousands of scientific articles, clinical trials, guidelines, and biochemical reports. Clinicians and researchers face the dual challenge of locating this information and judging its reliability for pediatric use. The complexity and unstructured nature of biomedical literature can slow the path from diagnosis to action.

To support rapid genome sequencing programs, newborn screening, and acute pediatric decision making, RCIGM required a scientifically rigorous methodology that could transform dispersed data into structured, actionable, and clinically ready intelligence.

# The Rancho Solution

Rancho applied a proven, high-precision scientific data methodology specifically designed for complex biomedical evidence. The approach combines deep domain expertise with structured governance, reproducible workflows, and multi-level review.

**Comprehensive Knowledge Mining**

- Systematic capture of treatment evidence across PubMed, conference abstracts, clinical trials, guidelines, and industry publications.
- Coverage of 700 gene disease combinations with full traceability to source literature.

**Expert Scientific Curation**

- Evaluation of treatment intent, pediatric appropriateness, contraindications, precautions, and estimated time to treat.
- Classification of treatments as curative, effective or ameliorative, still in trials or unknown, or contraindicated.
- Direct alignment with target gene variant evidence.

**Rigorous Quality, Governance, and Reviewer Alignment**

- Independent scientific review of every curated treatment entry. A governance team validating evidence integrity, data completeness, and consistency across genes and disorders.
- Structured workflows in RedCAP to record edits, compare reviewer assessments, and finalize clinical recommendations.
- Annual review cycles that ensure the data evolves with scientific knowledge.

**Ready for Clinical Decision Support**

- Rancho's harmonized knowledge base integrates seamlessly into the BeginNGS platform, where clinicians can access treatment insights within minutes after diagnosis.

# Impact

From Hours to Minutes - Quantifiable Gains in Treatment Readiness. The RCIGM and Rancho collaboration created a reliable and scalable infrastructure for rapid treatment decision making.

**585**
disorders reviewed for ICU use

**455**
disorders reviewed for newborn screening

**1040**
disease gene pairs curated and available for clinical use.

All 28 positive cases identified in the sequencing program benefited directly from curated treatment options.

18003 total treatments identified, with 2043 retained after expert review (11.3%).

A Phase 3 clinical trial of more than 750 children is underway, with 3.7% diagnosed with rare diseases supported by curated insights.

1208 of 10830 ICU treatments were approved (11.6%).

8357 of 75323 newborn screening treatments were approved (11.09%).

## Curation time per gene disease pair dropped from 15 hours in 2020 to under 6 hours in 2025.

## The Downstream Value of High-Quality Curated Evidence

- Clinicians receive treatment guidance in minutes, not hours.
- Newborns gain faster access to evidence-based interventions.
- Evidence quality and consistency improve across clinical teams.
- The dataset supports expansion into large scale newborn screening programs.
- Rancho created a reusable evidence framework valuable across many therapeutic areas.
- The methodology is directly applicable to biopharma use cases in real world evidence, translational research, safety, and AI model training.


## Scaling Treatment Intelligence Across Leading Health Systems

Multiple institutions across the GTRx and BeginNGS networks, including Balboa Naval Medical Center, Grossmont Hospital, Loma Linda Medical Center, Rancho Springs Medical Center, Scripps Medical Network, and UCSD Medical Network, rely on this curated dataset to support clinical decision making.

## A Blueprint for Biopharma Evidence Generation and AI Readiness

Biopharma organizations face similar challenges in synthesizing biomedical literature, clinical evidence, and mechanistic knowledge across therapeutic areas. Rancho's work with RCIGM demonstrates:

- The ability to convert unstructured data into high quality scientific intelligence.
- A scalable methodology that supports precision medicine and rare disease programs.
- A foundation for training AI, LLMs, and agentic decision systems with trusted data.
- A repeatable framework for evidence generation across discovery, translational research, and clinical development.

Rancho delivers the scientific depth and data discipline required to accelerate insights, reduce uncertainty, and support more confident, data driven decisions across the R&D lifecycle.

## Conclusion

By combining genomic expertise, scientific curation, and rigorous data engineering, Rancho BioSciences enabled RCIGM to transform the speed and quality of rare disease treatment decisions. This collaboration showcases how high integrity data and expert curation can unlock meaningful clinical impact and serves as a model for biopharma organizations seeking to scale evidence development, improve decision quality, and prepare their R&D ecosystems for the next generation of AI powered scientific discovery.

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