

# Redefining Fabry Disease Care: Gene Therapy and Long-Acting ERT Prepare to Disrupt Status Quo | Competitive Intelligence

*Fabry disease treatment is advancing rapidly with gene therapies and next-gen ERTs promising durable efficacy, reduced burden, and broader mutation coverage.*

AUSTIN, TX, UNITED STATES, June 15, 2025 /EINPresswire.com/ -- [Fabry Disease](#), a rare, X-linked lysosomal storage disorder, is entering a new era of treatment innovation. Caused by a deficiency of the enzyme  $\alpha$ -galactosidase A ( $\alpha$ -Gal A), the disease leads to toxic accumulation of glycosphingolipids-particularly globotriaosylceramide (GL-3)-in critical organs including the kidneys, heart, and nervous system. Over time, patients experience progressive multi-system complications, including renal failure, cardiac disease, and cerebrovascular events.

“

Emerging therapies in Fabry disease are tackling longstanding gaps in CNS access, convenience, and mutation coverage-reshaping what long-term care could look like.”

*DataM Intelligence*

with atypical or late-onset forms.

Current Therapies: A Crowded but Fragmented Market

As of 2025, Fabry disease treatment revolves around two major classes:



Download CI Sample Report:

<https://www.datamintelligence.com/strategic-insights/sample/fabry-disease>

Global Epidemiology and Diagnostic Landscape

Fabry disease affects approximately 1 in every 40,000 to 117,000 live births worldwide, though prevalence is likely underestimated due to underdiagnosis and variability in phenotype presentation. Advances in newborn screening and genetic testing are expected to reveal a higher prevalence in coming years, particularly among patients

### Enzyme Replacement Therapy (ERT):

- Fabrazyme® (Sanofi) remains the gold-standard ERT, approved globally and backed by over a decade of data. It holds ~53% market share and is widely used in the U.S. and Europe.
- Replagal® (Takeda) is an alternative ERT, primarily marketed in Europe and Japan, but not approved in the U.S.
- Elfabrio® (Chiesi/Protalix), a newer PEGylated enzyme, boasts a longer half-life and potentially better tissue uptake. It is beginning to gain traction as a switch therapy, especially for patients seeking reduced infusion burden.

### Pharmacological Chaperone:

- Galafold® (Amicus), the only oral treatment, selectively stabilizes endogenous  $\alpha$ -Gal A in patients with amenable GLA mutations (~35–50% of Fabry patients). Its oral dosing and convenience appeal to many, though its mutation-specific applicability limits broader use.

### Market Dynamics: Key Trends & Challenges

Despite four approved therapies, Fabry disease care remains challenged by treatment burden, delayed onset, incomplete symptom control, and limited CNS access.

### Key Market Observations:

Current enzyme therapies are administered intravenously every two weeks, leading to high treatment burden and compliance issues.

- Galafold, while convenient as an oral agent, only serves a subset of patients with specific mutations.
- Central nervous system (CNS) symptoms are not addressed effectively due to poor blood-brain barrier penetration of current therapies.
- The annual treatment cost can exceed \$500,000, creating pressure for more sustainable, cost-effective options.

### Pipeline Outlook: Curative Therapies on the Horizon

A strong pipeline of gene therapies and advanced biologics is aiming to transform the Fabry disease treatment paradigm. These candidates promise:

- Durable, potentially curative enzyme expression via gene therapy.
- Broader mutation coverage, including patients ineligible for chaperone therapy.
- Better tissue and CNS penetration to tackle neurologic manifestations.
- Reduced immunogenicity and adverse effects compared to current IV-administered biologics.
- Simplified regimens with monthly, quarterly, or single-administration dosing.

### Target Opportunity Profile (TOP): Strategic Differentiators for Emerging Therapies

To effectively displace existing therapies like Fabrazyme, Replagal, and Galafold, emerging drugs must demonstrate several key advantages:

- Mechanism of Action: Instead of providing exogenous enzymes (like ERT) or stabilizing mutant enzymes (like Galafold), future therapies should aim to deliver sustained, endogenous enzyme production-through gene therapy or next-gen biologics-with superior tissue distribution.
- Route of Administration: Most current therapies require lifelong biweekly IV infusions or regular oral dosing. Ideal emerging treatments would be less invasive-such as single IV doses, monthly infusions, or oral therapies with less frequent administration.
- Dosing Frequency: There is a significant opportunity for therapies with reduced dosing frequency. One-time gene therapies or infrequent maintenance regimens (monthly or quarterly) would drastically reduce treatment burden.
- Efficacy: While approved therapies can reduce GL-3 accumulation and slow organ damage, the bar is rising. Emerging therapies must demonstrate the ability to halt or even reverse renal and cardiac damage, and achieve sustained biomarker normalization.
- Onset of Action: Faster therapeutic onset and sustained outcomes are highly valued, especially in patients with progressive disease.
- Safety & Tolerability: Infusion reactions and anti-drug antibodies limit current ERTs. Next-gen options should offer lower immunogenicity and improved tolerability without off-target effects.
- Mutation Coverage: Galafold is mutation-specific, limiting its use. Future therapies should work across all genotypes, expanding access and simplifying treatment decisions.
- CNS Penetration: A major unmet need is CNS involvement. Therapies capable of crossing the blood-brain barrier could offer benefits in preventing stroke and cognitive impairment.
- Patient Convenience: Biweekly infusions disrupt patients' lives. Oral agents, home-based administration, or single-infusion gene therapy would be game-changers in convenience.
- Diagnostic Simplicity: Broadly applicable therapies with fewer diagnostic prerequisites (e.g., mutation testing for amenability) will see faster uptake and broader clinical adoption.
- Cost Effectiveness: Gene therapies and long-acting agents may come at a premium, but must show value through improved outcomes, reduced hospital utilization, and fewer cumulative lifetime doses.

### Strategic Insights

- Sanofi's market dominance with Fabrazyme is increasingly under pressure, especially as patients and providers seek more convenient or less invasive options.
- Gene therapies may reset the standard of care by offering long-term efficacy with a single administration. However, questions remain around durability, manufacturing scalability, and pricing.
- Chiesi's Elfabrio and Amicus' Galafold are well-positioned as patient-friendly alternatives, particularly in switch populations or for mutation-specific subgroups.
- Payer interest is growing in value-based reimbursement models, especially as high upfront costs of gene therapy are weighed against potential long-term healthcare savings.

Book Your Free CI Consultation Call: <https://www.datamintelligence.com/strategic-insights/ci/fabry-disease>

## About Our Fabry Disease Competitive Intelligence Report

Our CI report provides critical, forward-looking insights for strategic stakeholders in the Fabry disease ecosystem:

- Track real-time updates on clinical trials, readouts, and regulatory approvals
- Benchmark your drug against pipeline competitors on key dimensions like efficacy, safety, convenience, and commercial potential
- Understand KOL and physician sentiment regarding treatment gaps, unmet needs, and adoption barriers
- Identify M&A, in-licensing, and co-development opportunities in this high-stakes rare disease market
- Shape your clinical and commercial strategy with precision

### Read Related Ci Reports

1. [Spinal Muscular Atrophy Genetic Understanding Disease Modifying Therapies](#)
2. [Retinal Vein Occlusion | Competitive Intelligence](#)

Sai Kumar

DataM Intelligence 4market Research LLP

+1 877-441-4866

[email us here](#)

Visit us on social media:

[LinkedIn](#)

[X](#)

---

This press release can be viewed online at: <https://www.einpresswire.com/article/822400414>

EIN Presswire's priority is source transparency. We do not allow opaque clients, and our editors try to be careful about weeding out false and misleading content. As a user, if you see something we have missed, please do bring it to our attention. Your help is welcome. EIN Presswire, Everyone's Internet News Presswire™, tries to define some of the boundaries that are reasonable in today's world. Please see our Editorial Guidelines for more information.

© 1995-2025 Newsmatics Inc. All Right Reserved.