BioMarin Pharmaceutical Patient Finder Analysis

Ada Patient Finder v4 Suitability Assessment | March 11, 2026

Executive Summary

BioMarin Pharmaceutical operates a portfolio of 10 approved rare disease therapies generating **$3.22B in FY2025 revenue**. This analysis evaluates Patient Finder suitability across the complete portfolio following the December 2025 acquisition of Amicus Therapeutics for $4.8B.

TIER 1 OPPORTUNITIES

3

Combined 1% penetration: **$6.2M/year**

TOTAL ADDRESSABLE (USA)

14,951

Undiagnosed patients across portfolio

HIGHEST FIT SCORE

9.0

Galafold (Fabry disease) - 10,000 US addressable

Portfolio Overview: Prioritized Opportunities

Drug	Indication	Tier	Fit Score	USA Addressable	Ada Revenue (1%)	Ada Revenue (5%)
Galafold (migalastat)	Fabry Disease	TIER 1	**9.0**	10,000	$2,050,000	$10,240,000
TOTAL TIER 1+2 OPPORTUNITY				**14,498**	**$3,596,000**	**$17,969,750**

Drug	Indication	Tier	Fit Score	USA Addressable	Ada Revenue (1%)	Ada Revenue (5%)
Pombiliti + Opfolda	Pompe Disease (LOPD)	TIER 1	**8.0**	4,200	$1,470,000	$7,350,000
Voxzogo (vosoritide)	Achondroplasia	TIER 1	**7.5**	280	$67,200	$336,000
Brineura (cerliponase alfa)	CLN2 Disease (Batten)	TIER 2	**6.0**	18	$8,800	$43,750
Palynziq (pegvaliase)	PKU (Phenylketonuria)	TIER 3	**4.5**	360*	$69,100	$345,500
Vimizim (elosulfase alfa)	MPS IVA (Morquio A)	TIER 3	**4.5**	55	$22,000	$110,000
ALDURAZYME (laronidase)	MPS I	TIER 3	**5.0**	38	$9,400	$46,950
Naglazyme (galsulfase)	MPS VI	NO	**3.5**	29	$6,400	$31,800
Kuvan (sapropterin)	PKU	NO	**N/A**	Generic competition (patent expired)		
Roctavian	Hemophilia A	NO	**N/A**	Being divested by BioMarin		
TOTAL TIER 1+2 OPPORTUNITY				**14,498**	**$3,596,000**	**$17,969,750**

**Palynziq: Primarily poorly controlled diagnosed patients (NBS covers >95%), not truly undiagnosed*

Tier 1: High-Priority Opportunities

1. Galafold (migalastat) - Fabry Disease

TIER 1 **Fit Score: 9.0**

Metric	Value
FY2024/2025 Revenue	$458M / $521.7M (+14% YoY)
Diagnostic Delay	**10-15 years**
Underdiagnosis Rate	**74% (!)**
USA Drug-Addressable	**10,000 patients**
Net Revenue per Patient	$204,750/year
Patent Expiry	2037-2038 (long exclusivity)
Strategic Priority	**$4.8B Amicus acquisition (Dec 2025)**

Opportunity Assessment

USA Opportunity: $204.75M total addressable | $2.05M at 1% | $10.24M at 5%

Combined (USA+DACH+ROW): $3.23M at 1% | $16.14M at 5%

Ada Surface Ability: 8/10 - Distinctive symptom cluster (neuropathic pain + hypohidrosis + GI issues + rash in pediatric/adolescent patients)

Company Motivation: 10/10 - $4.8B acquisition demonstrates extreme strategic priority

Pitch: "Fabry disease affects 37,000 symptomatic Americans, yet 74% remain undiagnosed—a staggering 27,800 patients. Diagnostic delays average 10-15 years as early symptoms (burning pain in hands/feet, inability to sweat, GI issues, distinctive rash) are dismissed as growing pains or IBS. BioMarin just acquired Galafold for $4.8B (Dec 2025), treating only 2,730 patients globally while ~10,000 drug-addressable Americans remain unfound. Ada's symptom checker can identify the distinctive pediatric/adolescent presentation, triggering genetic testing and capturing $205K annual revenue per patient. This represents BioMarin's largest undiagnosed patient pool and Ada's highest-value rare disease opportunity."

2. Pombiliti + Opfolda - Pompe Disease (Late-Onset)

TIER 1 **Fit Score: 8.0**

Metric	Value
FY2025 Revenue (9mo)	$77.5M (+61.5% YoY)
Diagnostic Delay	**12 years (median 144 months)**
Underdiagnosis Rate	**60-70%**
USA Drug-Addressable	**4,200 patients**
Net Revenue per Patient	~$350,000/year (estimated)
Misdiagnosis Rate	34% initially misdiagnosed
Strategic Priority	**$4.8B Amicus acquisition (Dec 2025)**

Opportunity Assessment

USA Opportunity: $147M total addressable | $1.47M at 1% | $7.35M at 5%

Combined (USA+DACH+ROW): $2.38M at 1% | $11.92M at 5%

Ada Surface Ability: 7/10 - Proximal weakness + respiratory insufficiency + elevated CK

Company Motivation: 9/10 - Included in $4.8B acquisition, 61.5% YoY growth

Pitch: "Pompe disease affects 10,000 Americans with late-onset muscle weakness, yet diagnostic delays average 12 years—leaving 4,200+ undiagnosed as they're misdiagnosed with generic 'muscular dystrophy.' Proximal weakness (trouble climbing stairs, rising from chairs) plus respiratory issues in 20-50 year-olds is a distinctive pattern. BioMarin acquired Pombiliti+Opfolda in a $4.8B deal (Dec 2025), and the combination therapy beat Sanofi's standard ERT in clinical trials. With only ~4,000 treated globally vs. 8,000+ diagnosed and another 15,000 undiagnosed worldwide, patient finding is critical."

3. Voxzogo (vosoritide) - Achondroplasia

TIER 1 **Fit Score: 7.5**

Metric	Value
FY2024/2025 Revenue	$735M / $927M (+26% YoY)
Market Position	**BioMarin's largest revenue product**
Diagnostic Delay	Minimal (typically diagnosed at birth)
Underdiagnosis Rate	<5% (highly visible skeletal dysplasia)
USA Drug-Addressable	**280 patients** (milder cases, late presenters)
Net Revenue per Patient	$240,000/year
Competitive Threat	**BridgeBio oral competitor (Phase 3 positive Feb 2026)**

Opportunity Assessment

USA Opportunity: $6.72M total addressable | $67K at 1% | $336K at 5%

Combined (USA+DACH+ROW): $620K at 1% | $3.1M at 5%

Ada Surface Ability: 8/10 - Clear symptoms (short stature, disproportion, large head)

Company Motivation: 7/10 - Largest product, but facing oral competition from 2027+

Pitch: "Achondroplasia affects 14,500 Americans, yet ~280 pediatric patients remain undiagnosed—particularly milder cases missed in early growth monitoring. With Voxzogo generating $927M and facing oral competition from BridgeBio by 2027, every early diagnosis matters. Ada's symptom checker can identify growth concerns, disproportionate limbs, and family history patterns to flag potential cases before growth plates close, capturing $240K annual revenue per patient found."

Tier 2: Pursue

4. Brineura (cerliponase alfa) - CLN2 Disease (Batten Disease)

TIER 2 **Fit Score: 6.0**

Metric	Value
Estimated Revenue	<$50M (ultra-rare)
Diagnostic Delay	**2.05 years (range 0.33-8 years)**
USA Drug-Addressable	**18 patients**
Net Revenue per Patient	$486,000/year
Recent Activity	Label expansion July 2024 (children <3 years)
Competitive Threat	Gene therapy competition looming (2027+)

Opportunity Assessment

USA Opportunity: $875K total addressable | $8.8K at 1% | $44K at 5%

Ada Surface Ability: 7/10 - Distinctive cluster (seizures + language regression at age 2-4)

Company Motivation: 6/10 - Recent expansion shows interest, but ultra-rare limits scale

Pitch: "CLN2 disease affects ~100 American children, yet 2-year diagnostic delays mean 18+ remain undiagnosed—often misdiagnosed as epilepsy or autism. Early symptoms (seizures + language regression at age 2-4) are distinctive but require high suspicion. With Brineura's recent expansion to infants and gene therapy competition emerging by 2027, capturing patients early is critical before irreversible neurodegeneration occurs."

Strategic Recommendations

1. Immediate High-Value Targets

Galafold and Pombiliti+Opfolda offer **$5.6M combined annual opportunity at 1% penetration, $28M at 5%**. Both have massive undiagnosed pools (10,000+ and 4,200+ respectively), long diagnostic delays (10-15 years and 12 years), and are BioMarin's newest assets from the $4.8B Amicus acquisition (Dec 2025).

2. Symptom Checker Development Priority

Build algorithms for:

- **Fabry:** Neuropathic pain + hypohidrosis + GI issues + rash (pediatric/adolescent presentation)
- **Pompe:** Proximal weakness + respiratory insufficiency + elevated CK (ages 20-50)
- **Achondroplasia:** Disproportionate short stature + skeletal features + growth concerns
- **CLN2:** Seizures + language regression (ages 2-4)

3. BioMarin Partnership Angle

The **$4.8B Amicus acquisition (Dec 2025)** demonstrates extreme patient finding need for Galafold and Pombiliti+Opfolda. Position Ada as the solution to unlock the **74% undiagnosed Fabry population** and **60% undiagnosed Pompe population**. This is pure market expansion, not patient recruitment.

4. Market Expansion Positioning

Frame as **"market expansion"** (BioMarin's language), not "patient finding." Fabry's 2,730 current patients vs. 15,000 US+DACH addressable represents **5x expansion potential**—purely through diagnosis, not market share gains.

5. Competitive Urgency

- **Voxzogo:** BridgeBio's oral alternative achieves Phase 3 approval by 2027+, creating urgency for early patient capture
- **Brineura:** Gene therapy competition post-2027 threatens market share
- **Galafold:** Long patent exclusivity (2037-2038) provides runway, but early patient starts maximize lifetime value

Key Insights

Insight Category	Finding

Largest Opportunity	Fabry disease: 10,000 US drug-addressable undiagnosed patients, 74% underdiagnosis rate, 10-15 year diagnostic delays. $204.75M total addressable market for Ada.
Highest Strategic Priority	$4.8B Amicus acquisition (Dec 2025) makes Galafold and Pombiliti+Opfolda BioMarin's top growth drivers. Combined 16,310 addressable undiagnosed patients globally.
Best Symptom Profile	Fabry (neuropathic pain + hypohidrosis + GI + rash in youth) and CLN2 (seizures + language regression age 2-4) offer highly distinctive symptom clusters for Ada's assessment tool.
Competitive Dynamics	Voxzogo faces oral competition from BridgeBio (2027+); Brineura faces gene therapy threats (2027+). Early patient capture critical before market share erosion.
Ultra-Rare Limitations	MPS conditions (IVA, VI, I) have <100 US drug-addressable patients each. Small pools and mature products limit Patient Finder ROI despite high per-patient fees.
Newborn Screening Impact	PKU (>95% NBS coverage) and MPS I (45+ states) have limited truly undiagnosed populations. Better fit for treatment optimization vs. patient finding.

Tier 3 & Excluded Assets

Drug	Tier/Rationale	Key Issue
Palynziq (PKU)	Tier 3 (Fit 4.5)	NBS covers >95% of PKU cases. Addressable pool is poorly controlled diagnosed patients, not truly undiagnosed. Better fit: treatment optimization finder.
Vimizim (MPS IVA)	Tier 3 (Fit 4.5)	Ultra-rare (55 US addressable), mature product with declining strategic priority. 3-year delays indicate missed diagnoses, but small pool limits ROI.
ALDURAZYME (MPS I)	Tier 3 (Fit 5.0)	Newborn screening reducing undiagnosed pool. Small addressable (38 US patients). Sanofi partnership limits BioMarin direct control.
Naglazyme (MPS VI)	NO (Fit 3.5)	Ultra-rare (29 US addressable), mature, patent-expired, declining priority. Among the smallest patient pools in BioMarin's portfolio.
Kuvan (PKU)	NO	Generic competition (patent expired 2024-2026), declining revenues. Strategic focus shifted to Palynziq.

Roctavian (Hemophilia A)	NO	Being divested by BioMarin (2025-2026) after commercial failure. $119M write-down. Not viable target.

BioMarin Patient Finder Analysis - Ada Cockpit - March 2026

Prepared by Ada Health Research Team | For internal strategic use

This analysis evaluated 10 approved BioMarin drugs across USA, DACH, and ROW markets. Scoring methodology: Fit Score = (Ada Surface Ability × 0.5) + (Company Motivation × 0.3) + (Market Size × 0.2). All revenue figures in USD. Sources cited throughout full markdown report.