Updated: 08/2026
DMMA Approved: 08/2026

Request for Prior Authorization for Fabry Disease Medications
Website Form – www.highmarkhealthoptions.com
Submit request via: Fax - 1-855-476-4158

All requests for Fabry Disease Medications require a Prior Authorization and will be screened for medical necessity and appropriateness using the criteria listed below.

Fabry Disease Medications Prior Authorization Criteria:

Fabry Disease Medications include Galafold (migalastat), Fabrazyme (agalsidase beta), or Elfabrio (pegunigalsidase alfa-iwxj). New products with this classification will require the same documentation.

For all requests for Fabry Disease Medications all of the following criteria must be met:

Coverage may be provided with a diagnosis of Fabry Disease and the following criteria is met:

- Diagnosis has been confirmed by biochemical/genetic confirmation by ONE of the following:
  - α-galactosidase A (α-Gal A) activity in plasma, isolated leukocytes, and/or cultured cells.
  - Plasma or urinary globotriaosylceramide (Gb3/GL-3) or globotriaosylsphingosine (lyso-Gb3).
  - Detection of pathogenic mutations in the GALA/GLA gene by molecular genetic testing.
- Documentation the member is ONE of the following:
  - Symptomatic (i.e. intermittent episodes of burning pain in the extremities (acroparesthesias); cutaneous vascular lesions (angiokeratomas); diminished perspiration (hypo- or anhidrosis); characteristic corneal and lenticular opacities; abdominal pain, nausea, and/or diarrhea of unknown etiology in young adulthood; left ventricular hypertrophy (LVH) or hypertrophic cardiomyopathy of unknown etiology, particularly in young adults; arrhythmias of unknown etiology, particularly in young adults; stroke of unknown etiology at any age; chronic kidney disease (CKD) and/or proteinuria of unknown etiology; multiple renal sinus cysts discovered incidentally)
  - Asymptomatic with all of the following:
    - Assigned male at birth
    - Have classic Fabry mutations
  - Documentation of biopsy evidence indicating initiation of enzyme replacement therapy is medically necessary.
- Medication must be prescribed by or in consultation with a geneticist, dermatologist, neurologist, nephrologist, rheumatologist, or cardiologist.
- The requested dose and frequency is in accordance with FDA-approved labeling, nationally recognized compendia, and/or evidence-based practice guidelines.

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For all requests for Fabrazyme (agalsidase beta) or Elfabrio (pegunigalsidase alfa-iwxj) all of the following criteria must be met:

- **Initial Duration of Approval:** 12 months
- **Reauthorization criteria**
  - Chart documentation demonstrating clinical benefit and tolerance to the requested medication
- **Reauthorization Duration of approval:** 12 months

For all requests for Galafold (migalastat) all of the following criteria must be met:

- Member must have amenable GLA variant that is interpreted by a clinical genetics professional as causing Fabry disease (pathogenic, likely pathogenic) in the clinical context of the patient. (see attachment 1)
- Exclusion criteria
  - Member must not have severe renal impairment (eGFR <30 mL/minute/1.73 m^2)
  - Member must not have end-stage renal disease requiring dialysis
- **Initial Duration of Approval:** 6 months
- **Reauthorization criteria**
  - Chart documentation demonstrating clinical benefit and tolerance to Galafold.
- **Reauthorization Duration of Approval:** 12 months

Coverage may be provided for any non-FDA labeled indication if it is determined that the use is a medically accepted indication supported by nationally recognized pharmacy compendia or peer-reviewed medical literature for treatment of the diagnosis(es) for which it is prescribed. These requests will be reviewed on a case by case basis to determine medical necessity.

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FABRY DISEASE MEDICATIONS PRIOR AUTHORIZATION FORM

Please complete and fax all requested information below including any progress notes, laboratory test results, or chart documentation as applicable to Highmark Health Options Pharmacy Services. **FAX:** (855) 476-4158
If needed, you may call to speak to a Pharmacy Services Representative.
PHONE: (844) 325-6251 Monday through Friday 8:00am to 7:00pm

PROVIDER INFORMATION

Requesting Provider:	NPI:
Provider Specialty:	Office Contact:
Office Address:	Office Phone:
	Office Fax:

MEMBER INFORMATION

Member Name:	DOB:	
Health Options ID:	Member weight:	Height:

REQUESTED DRUG INFORMATION

Medication:	Strength:	
Directions:	Quantity:	Refills:
Is the member currently receiving requested medication? ☐ Yes ☐ No	Date Medication Initiated:	

Is this medication being used for a chronic or long-term condition for which the medication may be necessary for the life of the patient? ☐ Yes ☐ No

Billing Information

This medication will be billed: ☐ at a pharmacy **OR**
☐ medically (if medically please provide a JCODE: _____

Place of Service: ☐ Hospital ☐ Provider's office ☐ Member's home ☐ Other

Place of Service Information

Name:	NPI:
Address:	Phone:

MEDICAL HISTORY (Complete for ALL requests)

Member's Diagnosis: ☐ Fabry Disease ☐ Other _____

For all medication requests:
How was the member's diagnosis confirmed? (please submit documentation and check one of the following)
☐ α-galactosidase A (α-Gal A) activity in plasma, isolated leukocytes, and/or cultured cells.
☐ Plasma or urinary globotriaosylceramide(Gb3/GL-3) or globotriaosylsphingosine (lyso-Gb3).
☐ Detection of pathogenic mutations in the GALA/GLA gene by molecular genetic testing.

Please select one of the following:
☐ The member is experiencing symptoms (please submit documentation)
☐ The member is asymptomatic and meets the following criteria:
  ☐ The member was assigned male at birth
  ☐ The member is 8 years of age and older
  ☐ The member has classic Fabry mutations
☐ The member had a biopsy that showed evidence indicating initiation of enzyme replacement therapy is medically necessary (please submit documentation)

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FABRY DISEASE MEDICATIONS
PRIOR AUTHORIZATION FORM (CONTINUED) – PAGE 2 OF 2

Please complete and fax all requested information below including any progress notes, laboratory test results, or chart documentation as applicable to Highmark Health Options Pharmacy Services. **FAX: (855) 476-4158**
If needed, you may call to speak to a Pharmacy Services Representative. **PHONE**: (844) 325-6251 Mon – Fri 8 am to 7 pm

MEMBER INFORMATION

Member Name:	DOB:	
Member ID:	Member weight:	Height:

MEDICAL HISTORY (Complete for ALL requests)

For requests for Galafold (migalastat)
Does the member have amenable GLA variant that has been interpreted by a clinical genetics professional as causing Fabry disease (pathogenic, likely pathogenic) in the clinical context of the patient? ☐ Yes ☐ No

Please provide variant: _____

Does the member have severe renal impairment (eGFR <30 mL/minute/1.73 m^2)? ☐ Yes ☐ No

Does the member have end-stage renal disease requiring dialysis? ☐ Yes ☐ No

CURRENT or PREVIOUS THERAPY

Medication Name	Strength/ Frequency	Dates of Therapy	Status (Discontinued & Why/Current)

REAUTHORIZATION

Has the member tolerated treatment and experienced clinical benefit? ☐ Yes ☐ No
Please describe:

SUPPORTING INFORMATION or CLINICAL RATIONALE

Prescribing Provider Signature	Date

Attachment: 1

Table 2: Amenable *GLA* Variants Based on the In Vitro Assay

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.7C>G	c.C7G	p.(L3V)	p.(Leu3Val)
c.8T>C	c.T8C	p.(L3P)	p.(Leu3Pro)
c.[11G>T; 620A>C]	c.G11T/A620C	p.(R4M/Y207S)	p.(Arg4Met/Tyr207Ser)
c.37G>A	c.G37A	p.(A13T)	p.(Ala13Thr)
c.37G>C	c.G37C	p.(A13P)	p.(Ala13Pro)
c.43G>A	c.G43A	p.(A15T)	p.(Ala15Thr)
c.44C>G	c.C44G	p.(A15G)	p.(Ala15Gly)
c.53T>G	c.T53G	p.(F18C)	p.(Phe18Cys)
c.58G>C	c.G58C	p.(A20P)	p.(Ala20Pro)

Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.59C>A	c.C59A	p.(A20D)	p.(Ala20Asp)
c.65T>G	c.T65G	p.(V22G)	p.(Val22Gly)
c.70T>C or c.70T>A	c.T70C or c.T70A	p.(W24R)	p.(Trp24Arg)
c.70T>G	c.T70G	p.(W24G)	p.(Trp24Gly)
c.72G>C or c.72G>T	c.G72C or c.G72T	p.(W24C)	p.(Trp24Cys)
c.95T>C	c.T95C	p.(L32P)	p.(Leu32Pro)
c.97G>T	c.G97T	p.(D33Y)	p.(Asp33Tyr)
c.98A>G	c.A98G	p.(D33G)	p.(Asp33Gly)
c.100A>C	c.A100C	p.(N34H)	p.(Asn34His)
c.100A>G	c.A100G	p.(N34D)	p.(Asn34Asp)
c.101A>C	c.A101C	p.(N34T)	p.(Asn34Thr)
c.101A>G	c.A101G	p.(N34S)	p.(Asn34Ser)
c.102T>G or c.102T>A	c.T102G or c.T102A	p.(N34K)	p.(Asn34Lys)
c.103G>C or c.103G>A	c.G103C or c.G103A	p.(G35R)	p.(Gly35Arg)
c.104G>A	c.G104A	p.(G35E)	p.(Gly35Glu)
c.104G>T	c.G104T	p.(G35V)	p.(Gly35Val)
c.107T>C	c.T107C	p.(L36S)	p.(Leu36Ser)
c.107T>G	c.T107G	p.(L36W)	p.(Leu36Trp)
c.108G>C or c.108G>T	c.G108C or c.G108T	p.(L36F)	p.(Leu36Phe)
c.109G>A	c.G109A	p.(A37T)	p.(Ala37Thr)
c.110C>T	c.C110T	p.(A37V)	p.(Ala37Val)
c.122C>T	c.C122T	p.(T41I)	p.(Thr41Ile)
c.124A>C or c.124A>T	c.A124C or c.A124T	p.(M42L)	p.(Met42Leu)
c.124A>G	c.A124G	p.(M42V)	p.(Met42Val)
c.125T>A	c.T125A	p.(M42K)	p.(Met42Lys)
c.125T>C	c.T125C	p.(M42T)	p.(Met42Thr)
c.125T>G	c.T125G	p.(M42R)	p.(Met42Arg)
c.126G>A or c.126G>C or c.126G>T	c.G126A or c.G126C or c.G126T	p.(M42I)	p.(Met42Ile)
c.137A>C	c.A137C	p.(H46P)	p.(His46Pro)

Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.142G>C	c.G142C	p.(E48Q)	p.(Glu48Gln)
c.152T>A	c.T152A	p.(M51K)	p.(Met51Lys)
c.153G>A or c.153G>T or c.153G>C	c.G153A or c.G153T or c.G153C	p.(M51I)	p.(Met51Ile)
c.[157A>C; 158A>T]	c.A157C/A158T	p.(N53L)	p.(Asn53Leu)
c.157A>G	c.A157G	p.(N53D)	p.(Asn53Asp)
c.160C>T	c.C160T	p.(L54F)	p.(Leu54Phe)
c.161T>C	c.T161C	p.(L54P)	p.(Leu54Pro)
c.164A>G	c.A164G	p.(D55G)	p.(Asp55Gly)
c.164A>T	c.A164T	p.(D55V)	p.(Asp55Val)
c.[164A>T; 170A>T]	c.A164T/A170T	p.(D55V/Q57L)	p.(Asp55Val/Gln57Leu)
c.167G>A	c.G167A	p.(C56Y)	p.(Cys56Tyr)
c.167G>T	c.G167T	p.(C56F)	p.(Cys56Phe)
c.170A>T	c.A170T	p.(Q57L)	p.(Gln57Leu)
c.175G>A	c.G175A	p.(E59K)	p.(Glu59Lys)
c.178C>A	c.C178A	p.(P60T)	p.(Pro60Thr)
c.178C>T	c.C178T	p.(P60S)	p.(Pro60Ser)
c.179C>T	c.C179T	p.(P60L)	p.(Pro60Leu)
c.196G>A	c.G196A	p.(E66K)	p.(Glu66Lys)
c.197A>G	c.A197G	p.(E66G)	p.(Glu66Gly)
c.207C>A or c.207C>G	c.C207A or c.C207G	p.(F69L)	p.(Phe69Leu)
c.214A>G	c.A214G	p.(M72V)	p.(Met72Val)
c.216G>A or c.216G>T or c.216G>C	c.G216A or c.G216T or c.G216C	p.(M72I)	p.(Met72Ile)
c.218C>T	c.C218T	p.(A73V)	p.(Ala73Val)
c.227T>C	c.T227C	p.(M76T)	p.(Met76Thr)
c.239G>A	c.G239A	p.(G80D)	p.(Gly80Asp)
c.239G>T	c.G239T	p.(G80V)	p.(Gly80Val)
c.247G>A	c.G247A	p.(D83N)	p.(Asp83Asn)
c.253G>A	c.G253A	p.(G85S)	p.(Gly85Ser)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.[253G>A; 254G>A]	c.G253A/G254A	p.(G85N)	p.(Gly85Asn)
c.[253G>A; 254G>T; 255T>G]	c.G253A/G254T/T255G	p.(G85M)	p.(Gly85Met)
c.254G>A	c.G254A	p.(G85D)	p.(Gly85Asp)
c.261G>C or c.261G>T	c.G261C or c.G261T	p.(E87D)	p.(Glu87Asp)
c.265C>T	c.C265T	p.(L89F)	p.(Leu89Phe)
c.272T>C	c.T272C	p.(I91T)	p.(Ile91Thr)
c.288G>A or c.288G>T or c.288G>C	c.G288A or c.G288T or c.G288C	p.(M96I)	p.(Met96Ile)
c.289G>C	c.G289C	p.(A97P)	p.(Ala97Pro)
c.290C>T	c.C290T	p.(A97V)	p.(Ala97Val)
c.305C>T	c.C305T	p.(S102L)	p.(Ser102Leu)
c.311G>T	c.G311T	p.(G104V)	p.(Gly104Val)
c.316C>T	c.C316T	p.(L106F)	p.(Leu106Phe)
c.320A>G	c.A320G	p.(Q107R)	p.(Gln107Arg)
c.322G>A	c.G322A	p.(A108T)	p.(Ala108Thr)
c.326A>G	c.A326G	p.(D109G)	p.(Asp109Gly)
c.334C>G	c.C334G	p.(R112G)	p.(Arg112Gly)
c.335G>A	c.G335A	p.(R112H)	p.(Arg112His)
c.337T>A	c.T337A	p.(F113I)	p.(Phe113Ile)
c.337T>C or c.339T>A or c.339T>G	c.T337C or c.T339A or c.T339G	p.(F113L)	p.(Phe113Leu)
c.352C>T	c.C352T	p.(R118C)	p.(Arg118Cys)
c.361G>A	c.G361A	p.(A121T)	p.(Ala121Thr)
c.368A>G	c.A368G	p.(Y123C)	p.(Tyr123Cys)
c.373C>T	c.C373T	p.(H125Y)	p.(His125Tyr)
c.374A>T	c.A374T	p.(H125L)	p.(His125Leu)
c.376A>G	c.A376G	p.(S126G)	p.(Ser126Gly)
c.383G>A	c.G383A	p.(G128E)	p.(Gly128Glu)
c.399T>G	c.T399G	p.(I133M)	p.(Ile133Met)
c.404C>T	c.C404T	p.(A135V)	p.(Ala135Val)

Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.408T>A or c.408T>G	c.T408A or c.T408G	p.(D136E)	p.(Asp136Glu)
c.416A>G	c.A416G	p.(N139S)	p.(Asn139Ser)
c.419A>C	c.A419C	p.(K140T)	p.(Lys140Thr)
c.427G>A	c.G427A	p.(A143T)	p.(Ala143Thr)
c.431G>A	c.G431A	p.(G144D)	p.(Gly144Asp)
c.431G>T	c.G431T	p.(G144V)	p.(Gly144Val)
c.434T>C	c.T434C	p.(F145S)	p.(Phe145Ser)
c.436C>T	c.C436T	p.(P146S)	p.(Pro146Ser)
c.437C>G	c.C437G	p.(P146R)	p.(Pro146Arg)
c.454T>C	c.T454C	p.(Y152H)	p.(Tyr152His)
c.454T>G	c.T454G	p.(Y152D)	p.(Tyr152Asp)
c.455A>G	c.A455G	p.(Y152C)	p.(Tyr152Cys)
c.466G>A	c.G466A	p.(A156T)	p.(Ala156Thr)
c.466G>T	c.G466T	p.(A156S)	p.(Ala156Ser)
c.467C>T	c.C467T	p.(A156V)	p.(Ala156Val)
c.471G>C or c.471G>T	c.G471C or c.G471T	p.(Q157H)	p.(Gln157His)
c.484T>G	c.T484G	p.(W162G)	p.(Trp162Gly)
c.493G>C	c.G493C	p.(D165H)	p.(Asp165His)
c.494A>G	c.A494G	p.(D165G)	p.(Asp165Gly)
c.496_497delinsTC	c.496_497delinsTC	p.(L166S)	p.(Leu166Ser)
c.496C>G	c.C496G	p.(L166V)	p.(Leu166Val)
c.[496C>G; 497T>G]	c.C496G/T497G	p.(L166G)	p.(Leu166Gly)
c.499C>G	c.C499G	p.(L167V)	p.(Leu167Val)
c.506T>C	c.T506C	p.(F169S)	p.(Phe169Ser)
c.511G>A	c.G511A	p.(G171S)	p.(Gly171Ser)
c.520T>C	c.T520C	p.(C174R)	p.(Cys174Arg)
c.520T>G	c.T520G	p.(C174G)	p.(Cys174Gly)
c.525C>G or c.525C>A	c.C525G or c.C525A	p.(D175E)	p.(Asp175Glu)
c.539T>G	c.T539G	p.(L180W)	p.(Leu180Trp)
c.540G>C or c.540G>T	c.G540C or c.G540T	p.(L180F)	p.(Leu180Phe)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.548G>A	c.G548A	p.(G183D)	p.(Gly183Asp)
c.548G>C	c.G548C	p.(G183A)	p.(Gly183Ala)
c.550T>A	c.T550A	p.(Y184N)	p.(Tyr184Asn)
c.551A>G	c.A551G	p.(Y184C)	p.(Tyr184Cys)
c.553A>G	c.A553G	p.(K185E)	p.(Lys185Glu)
c.559_564dup	c.559_564dup	p.(M187_S188dup)	p.(Met187_Ser188dup)
c.559A>G	c.A559G	p.(M187V)	p.(Met187Val)
c.560T>C	c.T560C	p.(M187T)	p.(Met187Thr)
c.561G>T or c.561G>A or c.561G>C	c.G561T or c.G561A or c.G561C	p.(M187I)	p.(Met187Ile)
c.567G>C or c.567G>T	c.G567C or c.G567T	p.(L189F)	p.(Leu189Phe)
c.572T>A	c.T572A	p.(L191Q)	p.(Leu191Gln)
c.581C>T	c.C581T	p.(T194I)	p.(Thr194Ile)
c.584G>T	c.G584T	p.(G195V)	p.(Gly195Val)
c.586A>G	c.A586G	p.(R196G)	p.(Arg196Gly)
c.593T>C	c.T593C	p.(I198T)	p.(Ile198Thr)
c.595G>A	c.G595A	p.(V199M)	p.(Val199Met)
c.596T>C	c.T596C	p.(V199A)	p.(Val199Ala)
c.596T>G	c.T596G	p.(V199G)	p.(Val199Gly)
c.599A>G	c.A599G	p.(Y200C)	p.(Tyr200Cys)
c.602C>A	c.C602A	p.(S201Y)	p.(Ser201Tyr)
c.602C>T	c.C602T	p.(S201F)	p.(Ser201Phe)
c.608A>T	c.A608T	p.(E203V)	p.(Glu203Val)
c.609G>C or c.609G>T	c.G609C or c.G609T	p.(E203D)	p.(Glu203Asp)
c.611G>T	c.G611T	p.(W204L)	p.(Trp204Leu)
c.613C>A	c.C613A	p.(P205T)	p.(Pro205Thr)
c.613C>T	c.C613T	p.(P205S)	p.(Pro205Ser)
c.614C>T	c.C614T	p.(P205L)	p.(Pro205Leu)
c.619T>C	c.T619C	p.(Y207H)	p.(Tyr207His)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.620A>C	c.A620C	p.(Y207S)	p.(Tyr207Ser)
c.623T>G	c.T623G	p.(M208R)	p.(Met208Arg)
c.628C>T	c.C628T	p.(P210S)	p.(Pro210Ser)
c.629C>T	c.C629T	p.(P210L)	p.(Pro210Leu)
c.638A>G	c.A638G	p.(K213R)	p.(Lys213Arg)
c.638A>T	c.A638T	p.(K213M)	p.(Lys213Met)
c.640C>T	c.C640T	p.(P214S)	p.(Pro214Ser)
c.641C>T	c.C641T	p.(P214L)	p.(Pro214Leu)
c.643A>G	c.A643G	p.(N215D)	p.(Asn215Asp)
c.644A>G	c.A644G	p.(N215S)	p.(Asn215Ser)
c.[644A>G; 937G>T*]	c.A644G/G937T*	p.(N215S/D313Y*)	p.(Asn215Ser/Asp313Tyr*)
c.644A>T	c.A644T	p.(N215I)	p.(Asn215Ile)
c.646T>G	c.T646G	p.(Y216D)	p.(Tyr216Asp)
c.647A>G	c.A647G	p.(Y216C)	p.(Tyr216Cys)
c.655A>C	c.A655C	p.(I219L)	p.(Ile219Leu)
c.656T>A	c.T656A	p.(I219N)	p.(Ile219Asn)
c.656T>C	c.T656C	p.(I219T)	p.(Ile219Thr)
c.659G>A	c.G659A	p.(R220Q)	p.(Arg220Gln)
c.659G>C	c.G659C	p.(R220P)	p.(Arg220Pro)
c.662A>C	c.A662C	p.(Q221P)	p.(Gln221Pro)
c.671A>C	c.A671C	p.(N224T)	p.(Asn224Thr)
c.671A>G	c.A671G	p.(N224S)	p.(Asn224Ser)
c.673C>G	c.C673G	p.(H225D)	p.(His225Asp)
c.683A>G	c.A683G	p.(N228S)	p.(Asn228Ser)
c.687T>A or c.687T>G	c.T687A or c.T687G	p.(F229L)	p.(Phe229Leu)
c.695T>C	c.T695C	p.(I232T)	p.(Ile232Thr)
c.712A>G	c.A712G	p.(S238G)	p.(Ser238Gly)
c.713G>A	c.G713A	p.(S238N)	p.(Ser238Asn)
c.716T>C	c.T716C	p.(I239T)	p.(Ile239Thr)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.717A>G	c.A717G	p.(I239M)	p.(Ile239Met)
c.720G>C or c.720G>T	c.G720C or c.G720T	p.(K240N)	p.(Lys240Asn)
c.724A>G	c.A724G	p.(I242V)	p.(Ile242Val)
c.724A>T	c.A724T	p.(I242F)	p.(Ile242Phe)
c.725T>A	c.T725A	p.(I242N)	p.(Ile242Asn)
c.725T>C	c.T725C	p.(I242T)	p.(Ile242Thr)
c.728T>G	c.T728G	p.(L243W)	p.(Leu243Trp)
c.729G>C or c.729G>T	c.G729C or c.G729T	p.(L243F)	p.(Leu243Phe)
c.730G>A	c.G730A	p.(D244N)	p.(Asp244Asn)
c.730G>C	c.G730C	p.(D244H)	p.(Asp244His)
c.733T>G	c.T733G	p.(W245G)	p.(Trp245Gly)
c.740C>G	c.C740G	p.(S247C)	p.(Ser247Cys)
c.747C>G or c.747C>A	c.C747G or c.C747A	p.(N249K)	p.(Asn249Lys)
c.749A>C	c.A749C	p.(Q250P)	p.(Gln250Pro)
c.749A>G	c.A749G	p.(Q250R)	p.(Gln250Arg)
c.750G>C	c.G750C	p.(Q250H)	p.(Gln250His)
c.758T>C	c.T758C	p.(I253T)	p.(Ile253Thr)
c.758T>G	c.T758G	p.(I253S)	p.(Ile253Ser)
c.760-762delGTT or c.761-763del	c.760_762delGTT or c.761_763del	p.(V254del)	p.(Val254del)
c.769G>C	c.G769C	p.(A257P)	p.(Ala257Pro)
c.770C>G	c.C770G	p.(A257G)	p.(Ala257Gly)
c.770C>T	c.C770T	p.(A257V)	p.(Ala257Val)
c.772G>C or c.772G>A	c.G772C or c.G772A	p.(G258R)	p.(Gly258Arg)
c.773G>T	c.G773T	p.(G258V)	p.(Gly258Val)
c.776C>A	c.C776A	p.(P259Q)	p.(Pro259Gln)
c.776C>G	c.C776G	p.(P259R)	p.(Pro259Arg)
c.776C>T	c.C776T	p.(P259L)	p.(Pro259Leu)
c.779G>A	c.G779A	p.(G260E)	p.(Gly260Glu)
c.779G>C	c.G779C	p.(G260A)	p.(Gly260Ala)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.781G>A	c.G781A	p.(G261S)	p.(Gly261Ser)
c.781G>C	c.G781C	p.(G261R)	p.(Gly261Arg)
c.781G>T	c.G781T	p.(G261C)	p.(Gly261Cys)
c.788A>G	c.A788G	p.(N263S)	p.(Asn263Ser)
c.790G>T	c.G790T	p.(D264Y)	p.(Asp264Tyr)
c.794C>T	c.C794T	p.(P265L)	p.(Pro265Leu)
c.800T>C	c.T800C	p.(M267T)	p.(Met267Thr)
c.805G>A	c.G805A	p.(V269M)	p.(Val269Met)
c.806T>C	c.T806C	p.(V269A)	p.(Val269Ala)
c.809T>C	c.T809C	p.(I270T)	p.(Ile270Thr)
c.810T>G	c.T810G	p.(I270M)	p.(Ile270Met)
c.811G>A	c.G811A	p.(G271S)	p.(Gly271Ser)
c.[811G>A; 937G>T*]	c.G811A/G937T*	p.(G271S/D313Y*)	p.(Gly271Ser/Asp313Tyr*)
c.812G>A	c.G812A	p.(G271D)	p.(Gly271Asp)
c.823C>G	c.C823G	p.(L275V)	p.(Leu275Val)
c.827G>A	c.G827A	p.(S276N)	p.(Ser276Asn)
c.829T>G	c.T829G	p.(W277G)	p.(Trp277Gly)
c.831G>T or c.831G>C	c.G831T or c.G831C	p.(W277C)	p.(Trp277Cys)
c.832A>T	c.A832T	p.(N278Y)	p.(Asn278Tyr)
c.835C>G	c.C835G	p.(Q279E)	p.(Gln279Glu)
c.838C>A	c.C838A	p.(Q280K)	p.(Gln280Lys)
c.840A>T or c.840A>C	c.A840T or c.A840C	p.(Q280H)	p.(Gln280His)
c.844A>G	c.A844G	p.(T282A)	p.(Thr282Ala)
c.845C>T	c.C845T	p.(T282I)	p.(Thr282Ile)
c.850A>G	c.A850G	p.(M284V)	p.(Met284Val)
c.851T>C	c.T851C	p.(M284T)	p.(Met284Thr)
c.860G>T	c.G860T	p.(W287L)	p.(Trp287Leu)
c.862G>C	c.G862C	p.(A288P)	p.(Ala288Pro)
c.866T>G	c.T866G	p.(I289S)	p.(Ile289Ser)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.868A>C or c.868A>T	c.A868C or c.A868T	p.(M290L)	p.(Met290Leu)
c.869T>C	c.T869C	p.(M290T)	p.(Met290Thr)
c.870G>A or c.870G>C or c.870G>T	c.G870A or c.G870C or c.G870T	p.(M290I)	p.(Met290Ile)
c.871G>A	c.G871A	p.(A291T)	p.(Ala291Thr)
c.877C>A	c.C877A	p.(P293T)	p.(Pro293Thr)
c.881T>C	c.T881C	p.(L294S)	p.(Leu294Ser)
c.884T>G	c.T884G	p.(F295C)	p.(Phe295Cys)
c.886A>G	c.A886G	p.(M296V)	p.(Met296Val)
c.886A>T or c.886A>C	c.A886T or c.A886C	p.(M296L)	p.(Met296Leu)
c.887T>C	c.T887C	p.(M296T)	p.(Met296Thr)
c.888G>A or c.888G>T or c.888G>C	c.G888A or c.G888T or c.G888C	p.(M296I)	p.(Met296Ile)
c.893A>G	c.A893G	p.(N298S)	p.(Asn298Ser)
c.897C>G or c.897C>A	c.C897G or c.C897A	p.(D299E)	p.(Asp299Glu)
c.898C>T	c.C898T	p.(L300F)	p.(Leu300Phe)
c.899T>C	c.T899C	p.(L300P)	p.(Leu300Pro)
c.901C>G	c.C901G	p.(R301G)	p.(Arg301Gly)
c.902G>A	c.G902A	p.(R301Q)	p.(Arg301Gln)
c.902G>C	c.G902C	p.(R301P)	p.(Arg301Pro)
c.902G>T	c.G902T	p.(R301L)	p.(Arg301Leu)
c.907A>T	c.A907T	p.(I303F)	p.(Ile303Phe)
c.908T>A	c.T908A	p.(I303N)	p.(Ile303Asn)
c.911G>A	c.G911A	p.(S304N)	p.(Ser304Asn)
c.911G>C	c.G911C	p.(S304T)	p.(Ser304Thr)
c.919G>A	c.G919A	p.(A307T)	p.(Ala307Thr)
c.922A>G	c.A922G	p.(K308E)	p.(Lys308Glu)
c.924A>T or c.924A>C	c.A924T or c.A924C	p.(K308N)	p.(Lys308Asn)
c.925G>C	c.G925C	p.(A309P)	p.(Ala309Pro)
c.926C>T	c.C926T	p.(A309V)	p.(Ala309Val)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.928C>T	c.C928T	p.(L310F)	p.(Leu310Phe)
c.931C>G	c.C931G	p.(L311V)	p.(Leu311Val)
c.935A>G	c.A935G	p.(Q312R)	p.(Gln312Arg)
c.936G>T or c.936G>C	c.G936T or c.G936C	p.(Q312H)	p.(Gln312His)
c.937G>T*	c.G937T*	p.(D313Y*)	p.(Asp313Tyr*)
c.[937G>T*; 1232G>A]	c.G937T*/G1232A	p.(D313Y*/G411D)	p.(Asp313Tyr*/Gly411Asp)
c.938A>G	c.A938G	p.(D313G)	p.(Asp313Gly)
c.946G>A	c.G946A	p.(V316I)	p.(Val316Ile)
c.947T>G	c.T947G	p.(V316G)	p.(Val316Gly)
c.950T>C	c.T950C	p.(I317T)	p.(Ile317Thr)
c.955A>T	c.A955T	p.(I319F)	p.(Ile319Phe)
c.956T>C	c.T956C	p.(I319T)	p.(Ile319Thr)
c.958A>C	c.A958C	p.(N320H)	p.(Asn320His)
c.959A>T	c.A959T	p.(N320I)	p.(Asn320Ile)
c.962A>G	c.A962G	p.(Q321R)	p.(Gln321Arg)
c.962A>T	c.A962T	p.(Q321L)	p.(Gln321Leu)
c.963G>C or c.963G>T	c.G963C or c.G963T	p.(Q321H)	p.(Gln321His)
c.964G>A	c.G964A	p.(D322N)	p.(Asp322Asn)
c.964G>C	c.G964C	p.(D322H)	p.(Asp322His)
c.966C>A or c.966C>G	c.C966A or c.C966G	p.(D322E)	p.(Asp322Glu)
c.967C>A	c.C967A	p.(P323T)	p.(Pro323Thr)
c.968C>G	c.C968G	p.(P323R)	p.(Pro323Arg)
c.973G>A	c.G973A	p.(G325S)	p.(Gly325Ser)
c.973G>C	c.G973C	p.(G325R)	p.(Gly325Arg)
c.978G>C or c.978G>T	c.G978C or c.G978T	p.(K326N)	p.(Lys326Asn)
c.979C>G	c.C979G	p.(Q327E)	p.(Gln327Glu)
c.980A>T	c.A980T	p.(Q327L)	p.(Gln327Leu)
c.983G>C	c.G983C	p.(G328A)	p.(Gly328Ala)
c.989A>G	c.A989G	p.(Q330R)	p.(Gln330Arg)

Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.1001G>A	c.G1001A	p.(G334E)	p.(Gly334Glu)
c.1010T>C	c.T1010C	p.(F337S)	p.(Phe337Ser)
c.1012G>A	c.G1012A	p.(E338K)	p.(Glu338Lys)
c.1013A>T	c.A1013T	p.(E338V)	p.(Glu338Val)
c.1016T>A	c.T1016A	p.(V339E)	p.(Val339Glu)
c.1027C>A	c.C1027A	p.(P343T)	p.(Pro343Thr)
c.1028C>T	c.C1028T	p.(P343L)	p.(Pro343Leu)
c.1033T>C	c.T1033C	p.(S345P)	p.(Ser345Pro)
c.1046G>C	c.G1046C	p.(W349S)	p.(Trp349Ser)
c.1055C>G	c.C1055G	p.(A352G)	p.(Ala352Gly)
c.1055C>T	c.C1055T	p.(A352V)	p.(Ala352Val)
c.1061T>A	c.T1061A	p.(I354K)	p.(Ile354Lys)
c.1066C>G	c.C1066G	p.(R356G)	p.(Arg356Gly)
c.1066C>T	c.C1066T	p.(R356W)	p.(Arg356Trp)
c.1067G>A	c.G1067A	p.(R356Q)	p.(Arg356Gln)
c.1067G>C	c.G1067C	p.(R356P)	p.(Arg356Pro)
c.1072G>C	c.G1072C	p.(E358Q)	p.(Glu358Gln)
c.1073A>C	c.A1073C	p.(E358A)	p.(Glu358Ala)
c.1073A>G	c.A1073G	p.(E358G)	p.(Glu358Gly)
c.1074G>T or c.1074G>C	c.G1074T or c.G1074C	p.(E358D)	p.(Glu358Asp)
c.1076T>C	c.T1076C	p.(I359T)	p.(Ile359Thr)
c.1078G>A	c.G1078A	p.(G360S)	p.(Gly360Ser)
c.1078G>T	c.G1078T	p.(G360C)	p.(Gly360Cys)
c.1079G>A	c.G1079A	p.(G360D)	p.(Gly360Asp)
c.1082G>A	c.G1082A	p.(G361E)	p.(Gly361Glu)
c.1082G>C	c.G1082C	p.(G361A)	p.(Gly361Ala)
c.1084C>A	c.C1084A	p.(P362T)	p.(Pro362Thr)
c.1085C>T	c.C1085T	p.(P362L)	p.(Pro362Leu)
c.1087C>T	c.C1087T	p.(R363C)	p.(Arg363Cys)

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Table 2: Amenable *GLA* Variants Based on the In Vitro Assay (Continued)

DNA Change (Long)	DNA Change (Short)	Protein Change (1-letter Code)	Protein Change (3-letter Code)
c.1088G>A	c.G1088A	p.(R363H)	p.(Arg363His)
c.1102G>A	c.G1102A	p.(A368T)	p.(Ala368Thr)
c.1117G>A	c.G1117A	p.(G373S)	p.(Gly373Ser)
c.1124G>A	c.G1124A	p.(G375E)	p.(Gly375Glu)
c.1139C>T	c.C1139T	p.(P380L)	p.(Pro380Leu)
c.1153A>G	c.A1153G	p.(T385A)	p.(Tyr385Ala)
c.1168G>A	c.G1168A	p.(V390M)	p.(Val390Met)
c.1172A>C	c.A1172C	p.(K391T)	p.(Lys391Thr)
c.1184G>A	c.G1184A	p.(G395E)	p.(Gly395Glu)
c.1184G>C	c.G1184C	p.(G395A)	p.(Gly395Ala)
c.1192G>A	c.G1192A	p.(E398K)	p.(Glu398Lys)
c.1202_1203insGACTTC	c.1202_1203insGACTTC	p.(T400_S401dup)	p.(Thr400_Ser401dup)
c.1208T>C	c.T1208C	p.(L403S)	p.(Leu403Ser)
c.1225C>A	c.C1225A	p.(P409T)	p.(Pro409Thr)
c.1225C>G	c.C1225G	p.(P409A)	p.(Pro409Ala)
c.1225C>T	c.C1225T	p.(P409S)	p.(Pro409Ser)
c.1228A>G	c.A1228G	p.(T410A)	p.(Thr410Ala)
c.1229C>T	c.C1229T	p.(T410I)	p.(Thr410Ile)
c.1232G>A	c.G1232A	p.(G411D)	p.(Gly411Asp)
c.1234A>C	c.A1234C	p.(T412P)	p.(Thr412Pro)
c.1235C>A	c.C1235A	p.(T412N)	p.(Thr412Asn)
c.1253A>G	c.A1253G	p.(E418G)	p.(Glu418Gly)
c.1261A>G	c.A1261G	p.(M421V)	p.(Met421Val)

* Based on available published data, the *GLA* variant c.937G>T, (p.(D313Y)) is considered benign (not causing Fabry disease). Consultation with a clinical genetics professional is strongly recommended in patients with Fabry disease who have this *GLA* variant as additional evaluations may be indicated.

If a *GLA* variant does not appear in Table 2, it is either non-amenable (if tested) or has not been tested for in vitro amenability. For further information, please contact Amicus Medical Information at 1-877-4AMICUS or medinfousa@amicusrx.com.