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**Request for Prior Authorization for Fabry Disease Medications**

Website Form – [www.highmarkhealthoptions.com](http://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:
  - $\alpha$ -galactosidase A ( $\alpha$ -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.



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- The requested dose and frequency is in accordance with FDA-approved labeling, nationally recognized compendia, and/or evidence-based practice guidelines.

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<sup>2</sup>)
  - 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? <input type="checkbox"/> Yes <input type="checkbox"/> 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? <input type="checkbox"/> Yes <input type="checkbox"/> 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)

- $\alpha$ -galactosidase A ( $\alpha$ -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)

**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<sup>2</sup>)?  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.(F113D)	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](mailto:medinfousa@amicusrx.com).