

# ガラフォルド®に反応性のある GLA変異一覧

ガラフォルドは「ミガーラスタットに反応性のあるGLA遺伝子変異を伴うファブリー病」に対して効能・効果を有しています。

ガラフォルドによる治療に対して反応性があると考えられるファブリー病患者を特定するために、医薬品の安全性に関する非臨床試験の実施の基準 (GLP) に準拠したガラフォルド反応性確認試験 (ヒト胎児腎細胞アッセイ系) が開発され、臨床試験で検証されています。

以下の表は、ガラフォルドに反応性があると分類されているGLA遺伝子変異 (現時点で理論上想定されるすべての変異) を示しています。ガラフォルドに反応性のある変異及び反応性のない変異、双方についての一覧表は、以下のウェブサイトに掲載しています：[www.galafoldamenabilitytable.com](http://www.galafoldamenabilitytable.com)

※本資料はファブリー病の確定診断を目的とした用途にはご使用できません。GLA遺伝子変異の病原性の判定に関しては、専門医による評価をお受けすることを推奨いたします。

「効能又は効果」、「用法及び用量」、「禁忌を含む注意事項等情報」等につきましては、裏表紙のDIをご参照ください。



ファブリー病治療剤 処方箋医薬品<sup>注)</sup>

薬価基準収載

**ガラフォルド® カプセル123mg**  
GALAFOLD® Capsules 123mg (ミガーラスタット塩酸塩)

注) 注意-医師等の処方箋により使用すること

## 2. 禁忌 (次の患者には投与しないこと)

本剤の成分に対し過敏症の既往歴のある患者

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塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.7C>G	c.C7G	L3V	c.40C>T	c.C40T	L14F
c.8T>C	c.T8C	L3P	c.41T>A	c.T41A	L14H
c.[11G>T; 620A>C]	c.G11T/A620C	R4M/Y2075*	c.43G>A	c.G43A	A15T
c.13A>G	c.A13G	N5D	c.44C>G	c.C44G	A15G
c.15C>G	c.C15G	N5K	c.49C>A	c.C49A	R17S
c.16C>A	c.C16A	P6T	c.49C>G	c.C49G	R17G
c.16C>T	c.C16T	P6S	c.49C>T	c.C49T	R17C
c.17C>A	c.C17A	P6Q	c.50G>A	c.G50A	R17H
c.17C>G	c.C17G	P6R	c.50G>C	c.G50C	R17P
c.17C>T	c.C17T	P6L	c.52T>A	c.T52A	F18I
c.19G>A	c.G19A	E7K	c.53T>G	c.T53G	F18C
c.20A>T	c.A20T	E7V	c.54C>G	c.C54G	F18L
c.21A>T	c.A21T	E7D	c.58G>C	c.G58C	A20P
c.22C>A	c.C22A	L8I	c.59C>A	c.C59A	A20D
c.23T>A	c.T23A	L8Q	c.59C>G	c.C59G	A20G
c.23T>C	c.T23C	L8P	c.62T>A	c.T62A	L21H
c.25C>T	c.C25T	H9Y	c.64G>A	c.G64A	V22I
c.26A>G	c.A26G	H9R	c.64G>C	c.G64C	V22L
c.26A>T	c.A26T	H9L	c.64G>T	c.G64T	V22F
c.27T>A	c.T27A	H9Q	c.65T>C	c.T65C	V22A
c.28C>A	c.C28A	L10M	c.65T>G	c.T65G	V22G
c.28C>G	c.C28G	L10V	c.67T>A	c.T67A	S23T
c.29T>A	c.T29A	L10Q	c.67T>C	c.T67C	S23P
c.29T>C	c.T29C	L10P	c.[70T>A;1255A>G]	c.T70A/A1255G	W24R/N419D
c.29T>G	c.T29G	L10R	c.70T>C または c.70T>A	c.T70C または c.T70A	W24R
c.31G>A	c.G31A	G11S	c.70T>G	c.T70G	W24G
c.31G>C	c.G31C	G11R	c.71G>C	c.G71C	W24S
c.31G>T	c.G31T	G11C	c.72G>C または c.72G>T	c.G72C または c.G72T	W24C
c.32G>A	c.G32A	G11D	c.73G>C	c.G73C	D25H
c.32G>T	c.G32T	G11V	c.77T>A	c.T77A	I26N
c.34T>A	c.T34A	C12S	c.79C>A	c.C79A	P27T
c.34T>C	c.T34C	C12R	c.79C>G	c.C79G	P27A
c.34T>G	c.T34G	C12G	c.79C>T	c.C79T	P27S
c.35G>A	c.G35A	C12Y	c.80C>T	c.C80T	P27L
c.37G>A	c.G37A	A13T	c.82G>C	c.G82C	G28R
c.37G>C	c.G37C	A13P	c.82G>T	c.G82T	G28W
c.38C>A	c.C38A	A13E	c.83G>A	c.G83A	G28E
c.38C>G	c.C38G	A13G	c.85G>C	c.G85C	A29P
c.40C>G	c.C40G	L14V	c.86C>A	c.C86A	A29D

\*同一染色体上にある多重変異を表しています。

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塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.86C>G	c.C86G	A29G	c.116C>A	c.C116A	T39K
c.86C>T	c.C86T	A29V	c.116C>G	c.C116G	T39R
c.88A>G	c.A88G	R30G	c.116C>T	c.C116T	T39M
c.94C>A	c.C94A	L32M	c.121A>G	c.A121G	T41A
c.94C>G	c.C94G	L32V	c.122C>A	c.C122A	T41N
c.95T>A	c.T95A	L32Q	c.122C>G	c.C122G	T41S
c.95T>C	c.T95C	L32P	c.122C>T	c.C122T	T41I
c.95T>G	c.T95G	L32R	c.124A>C または c.124A>T	c.A124C または c.A124T	M42L
c.97G>C	c.G97C	D33H	c.124A>G	c.A124G	M42V
c.97G>T	c.G97T	D33Y	c.125T>A	c.T125A	M42K
c.98A>C	c.A98C	D33A	c.125T>C	c.T125C	M42T
c.98A>G	c.A98G	D33G	c.125T>G	c.T125G	M42R
c.98A>T	c.A98T	D33V	c.126G>A または c.126G>C または c.126G>T	c.G126A または c.G126C または c.G126T	M42I
c.99C>G	c.C99G	D33E	c.128G>C	c.G128C	G43A
c.100A>C	c.A100C	N34H	c.133C>A	c.C133A	L45M
c.100A>G	c.A100G	N34D	c.133C>G	c.C133G	L45V
c.101A>C	c.A101C	N34T	c.136C>A	c.C136A	H46N
c.101A>G	c.A101G	N34S	c.136C>G	c.C136G	H46D
c.102T>G または c.102T>A	c.T102G または c.T102A	N34K	c.137A>C	c.A137C	H46P
c.103G>C または c.103G>A	c.G103C または c.G103A	G35R	c.138C>G	c.C138G	H46Q
c.104G>A	c.G104A	G35E	c.142G>C	c.G142C	E48Q
c.104G>C	c.G104C	G35A	c.143A>C	c.A143C	E48A
c.104G>T	c.G104T	G35V	c.149T>A	c.T149A	F50Y
c.106T>A	c.T106A	L36M	c.151A>G	c.A151G	M51V
c.106T>G	c.T106G	L36V	c.152T>A	c.T152A	M51K
c.107T>C	c.T107C	L36S	c.152T>C	c.T152C	M51T
c.107T>G	c.T107G	L36W	c.152T>G	c.T152G	M51R
c.108G>C または c.108G>T	c.G108C または c.G108T	L36F	c.153G>A または c.153G>T または c.153G>C	c.G153A または c.G153T または c.G153C	M51I
c.109G>A	c.G109A	A37T	c.157A>C	c.A157C	N53H
c.109G>T	c.G109T	A37S	c.[157A>C; 158A>T]	c.A157C/A158T	N53L
c.110C>A	c.C110A	A37E	c.157A>G	c.A157G	N53D
c.110C>G	c.C110G	A37G	c.157A>T	c.A157T	N53Y
c.110C>T	c.C110T	A37V	c.158A>C	c.A158C	N53T
c.112A>G	c.A112G	R38G	c.158A>G	c.A158G	N53S
c.112A>T	c.A112T	R38W	c.158A>T	c.A158T	N53I
c.113G>T	c.G113T	R38M	c.159C>G または c.159C>A	c.C159G または c.C159A	N53K
c.114G>C	c.G114C	R38S	c.160C>G	c.C160G	L54V
c.115A>G	c.A115G	T39A			
c.115A>T	c.A115T	T39S			

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c.160C>T	c.C160T	L54F	c.193A>G	c.A193G	S65G
c.161T>A	c.T161A	L54H	c.193A>T	c.A193T	S65C
c.161T>C	c.T161C	L54P	c.195T>A	c.T195A	S65R
c.161T>G	c.T161G	L54R	c.196G>A	c.G196A	E66K
c.163G>C	c.G163C	D55H	c.197A>G	c.A197G	E66G
c.163G>T	c.G163T	D55Y	c.197A>T	c.A197T	E66V
c.164A>C	c.A164C	D55A	c.198G>C	c.G198C	E66D
c.164A>G	c.A164G	D55G	c.199A>C	c.A199C	K67Q
c.164A>T	c.A164T	D55V	c.199A>G	c.A199G	K67E
c.[164A>T; 170A>T]	c.A164T/A170T	D55V/Q57L*	c.200A>C	c.A200C	K67T
c.165C>G	c.C165G	D55E	c.200A>T	c.A200T	K67M
c.167G>A	c.G167A	C56Y	c.201G>C	c.G201C	K67N
c.167G>T	c.G167T	C56F	c.202C>A	c.C202A	L68I
c.168C>G	c.C168G	C56W	c.205T>A	c.T205A	F69I
c.170A>G	c.A170G	Q57R	c.206T>A	c.T206A	F69Y
c.170A>T	c.A170T	Q57L	c.207C>A または c.207C>G	c.C207A または c.C207G	F69L
c.172G>A	c.G172A	E58K	c.208A>T	c.A208T	M70L
c.175G>A	c.G175A	E59K	c.209T>A	c.T209A	M70K
c.175G>C	c.G175C	E59Q	c.209T>G	c.T209G	M70R
c.176A>C	c.A176C	E59A	c.210G>C	c.G210C	M70I
c.176A>G	c.A176G	E59G	c.211G>C	c.G211C	E71Q
c.176A>T	c.A176T	E59V	c.212A>C	c.A212C	E71A
c.177G>C	c.G177C	E59D	c.212A>G	c.A212G	E71G
c.178C>A	c.C178A	P60T	c.212A>T	c.A212T	E71V
c.178C>G	c.C178G	P60A	c.213G>C	c.G213C	E71D
c.178C>T	c.C178T	P60S	c.214A>G	c.A214G	M72V
c.179C>A	c.C179A	P60Q	c.214A>T	c.A214T	M72L
c.179C>G	c.C179G	P60R	c.215T>C	c.T215C	M72T
c.179C>T	c.C179T	P60L	c.216G>A または c.216G>T または c.216G>C	c.G216A または c.G216T または c.G216C	M72I
c.182A>T	c.A182T	D61V	c.217G>A	c.G217A	A73T
c.183T>A	c.T183A	D61E	c.217G>T	c.G217T	A73S
c.184_185insTAG	c.184_185insTAG	S62delinsLA	c.218C>T	c.C218T	A73V
c.184T>C	c.T184C	S62P	c.[218C>T;525C>G]	c.C218T/C525G	A73V/D175E
c.184T>G	c.T184G	S62A	c.220G>A	c.G220A	E74K
c.185C>A	c.C185A	S62Y	c.221A>G	c.A221G	E74G
c.185C>G	c.C185G	S62C	c.221A>T	c.A221T	E74V
c.185C>T	c.C185T	S62F	c.222G>C	c.G222C	E74D
c.190A>C	c.A190C	I64L	c.223C>T	c.C223T	L75F
c.190A>G	c.A190G	I64V			

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塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.224T>C	c.T224C	L75P	c.253G>C	c.G253C	G85R
c.226A>G	c.A226G	M76V	c.253G>T	c.G253T	G85C
c.227T>C	c.T227C	M76T	c.254G>A	c.G254A	G85D
c.229G>A	c.G229A	V77I	c.254G>C	c.G254C	G85A
c.229G>C	c.G229C	V77L	c.257A>T	c.A257T	Y86F
c.232T>C	c.T232C	S78P	c.260A>G	c.A260G	E87G
c.233C>T	c.C233T	S78L	c.261G>C または c.261G>T	c.G261C または c.G261T	E87D
c.235G>A	c.G235A	E79K	c.262T>A	c.T262A	Y88N
c.235G>C	c.G235C	E79Q	c.262T>C	c.T262C	Y88H
c.236A>C	c.A236C	E79A	c.263A>C	c.A263C	Y88S
c.236A>G	c.A236G	E79G	c.263A>G	c.A263G	Y88C
c.236A>T	c.A236T	E79V	c.265C>G	c.C265G	L89V
c.237A>T	c.A237T	E79D	c.265C>T	c.C265T	L89F
c.238G>A	c.G238A	G80S	c.271A>C	c.A271C	I91L
c.238G>T	c.G238T	G80C	c.271A>T	c.A271T	I91F
c.239G>A	c.G239A	G80D	c.272T>C	c.T272C	I91T
c.239G>C	c.G239C	G80A	c.272T>G	c.T272G	I91S
c.239G>T	c.G239T	G80V	c.273T>G	c.T273G	I91M
c.242G>T	c.G242T	W81L	c.286A>G	c.A286G	M96V
c.244A>G	c.A244G	K82E	c.286A>T	c.A286T	M96L
c.245A>C	c.A245C	K82T	c.287T>C	c.T287C	M96T
c.245A>G	c.A245G	K82R	c.288G>A または c.288G>T または c.288G>C	c.G288A または c.G288T または c.G288C	M96I
c.245A>T	c.A245T	K82M	c.289G>A	c.G289A	A97T
c.246G>C	c.G246C	K82N	c.289G>C	c.G289C	A97P
c.247G>A	c.G247A	D83N	c.289G>T	c.G289T	A97S
c.248A>C	c.A248C	D83A	c.290C>A	c.C290A	A97D
c.248A>G	c.A248G	D83G	c.290C>T	c.C290T	A97V
c.248A>T	c.A248T	D83V	c.293C>A	c.C293A	P98H
c.249T>A	c.T249A	D83E	c.293C>G	c.C293G	P98R
c.250G>A	c.G250A	A84T	c.293C>T	c.C293T	P98L
c.250G>C	c.G250C	A84P	c.295C>G	c.C295G	Q99E
c.250G>T	c.G250T	A84S	c.296A>C	c.A296C	Q99P
c.251C>A	c.C251A	A84E	c.296A>G	c.A296G	Q99R
c.251C>G	c.C251G	A84G	c.296A>T	c.A296T	Q99L
c.251C>T	c.C251T	A84V	c.301G>C	c.G301C	D101H
c.253G>A	c.G253A	G85S	c.302A>C	c.A302C	D101A
c.[253G>A; 254G>A]	c.G253A/G254A	G85N	c.302A>G	c.A302G	D101G
c.[253G>A; 254G>T; 255T>G]	c.G253A/G254T/T255G	G85M	c.302A>T	c.A302T	D101V

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c.303T>A	c.T303A	D101E	c.338T>A	c.T338A	F113Y
c.304T>A	c.T304A	S102T	c.341C>T	c.C341T	P114L
c.304T>C	c.T304C	S102P	c.343C>A	c.C343A	H115N
c.304T>G	c.T304G	S102A	c.343C>G	c.C343G	H115D
c.305C>T	c.C305T	S102L	c.346G>C	c.G346C	G116R
c.310G>A	c.G310A	G104S	c.350T>C	c.T350C	I117T
c.311G>A	c.G311A	G104D	c.351T>G	c.T351G	I117M
c.311G>C	c.G311C	G104A	c.352C>T	c.C352T	R118C
c.311G>T	c.G311T	G104V	c.361G>A	c.G361A	A121T
c.313A>G	c.A313G	R105G	c.362C>T	c.C362T	A121V
c.314G>A	c.G314A	R105K	c.367T>A	c.T367A	Y123N
c.314G>C	c.G314C	R105T	c.367T>G	c.T367G	Y123D
c.314G>T	c.G314T	R105I	c.368A>C	c.A368C	Y123S
c.316C>A	c.C316A	L106I	c.368A>G	c.A368G	Y123C
c.316C>G	c.C316G	L106V	c.368A>T	c.A368T	Y123F
c.316C>T	c.C316T	L106F	c.370G>A	c.G370A	V124I
c.317T>A	c.T317A	L106H	c.371T>G	c.T371G	V124G
c.317T>C	c.T317C	L106P	c.373C>A	c.C373A	H125N
c.319C>A	c.C319A	Q107K	c.373C>G	c.C373G	H125D
c.319C>G	c.C319G	Q107E	c.373C>T	c.C373T	H125Y
c.320A>G	c.A320G	Q107R	c.374A>G	c.A374G	H125R
c.321G>C	c.G321C	Q107H	c.374A>T	c.A374T	H125L
c.322G>A	c.G322A	A108T	c.376A>G	c.A376G	S126G
c.323C>A	c.C323A	A108E	c.376A>T	c.A376T	S126C
c.323C>T	c.C323T	A108V	c.377G>T	c.G377T	S126I
c.325G>A	c.G325A	D109N	c.379A>G	c.A379G	K127E
c.325G>C	c.G325C	D109H	c.383G>A	c.G383A	G128E
c.325G>T	c.G325T	D109Y	c.383G>C	c.G383C	G128A
c.326A>C	c.A326C	D109A	c.385C>G	c.C385G	L129V
c.326A>G	c.A326G	D109G	c.388A>C	c.A388C	K130Q
c.327C>G	c.C327G	D109E	c.389A>T	c.A389T	K130M
c.328C>A	c.C328A	P110T	c.390G>C	c.C390C	K130N
c.334C>G	c.C334G	R112G	c.391C>G	c.C391G	L131V
c.335G>A	c.G335A	R112H	c.397A>C	c.A397C	I133L
c.335G>T	c.G335T	R112L	c.397A>G	c.A397G	I133V
c.337T>A	c.T337A	F113I	c.397A>T	c.A397T	I133F
c.337T>C または c.339T>A または c.339T>G	c.T337C または c.T339A または c.T339G	F113L	c.398T>C	c.T398C	I133T
c.337T>G	c.T337G	F113V	c.399T>G	c.T399G	I133M
			c.[399T>G; 434T>C]	c.T399G/T434C	I133M/F145S

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.403G>A	c.G403A	A135T	c.434T>C	c.T434C	F145S
c.403G>T	c.G403T	A135S	c.434T>G	c.T434G	F145C
c.404C>A	c.C404A	A135E	c.435C>G	c.C435G	F145L
c.404C>G	c.C404G	A135G	c.436C>A	c.C436A	P146T
c.404C>T	c.C404T	A135V	c.436C>G	c.C436G	P146A
c.406G>A	c.G406A	D136N	c.436C>T	c.C436T	P146S
c.407A>C	c.A407C	D136A	c.437C>A	c.C437A	P146H
c.407A>T	c.A407T	D136V	c.437C>G	c.C437G	P146R
c.408T>A または c.408T>G	c.T408A または c.T408G	D136E	c.437C>T	c.C437T	P146L
c.409G>A	c.G409A	V137I	c.440G>C	c.G440C	G147A
c.409G>C	c.G409C	V137L	c.442A>G	c.A442G	S148G
c.410T>A	c.T410A	V137D	c.442A>T	c.A442T	S148C
c.410T>C	c.T410C	V137A	c.443G>C	c.G443C	S148T
c.410T>G	c.T410G	V137G	c.446T>G	c.T446G	F149C
c.413G>C	c.G413C	G138A	c.449G>A	c.G449A	G150E
c.415A>C	c.A415C	N139H	c.449G>T	c.G449T	G150V
c.415A>T	c.A415T	N139Y	c.451T>G	c.T451G	Y151D
c.416A>G	c.A416G	N139S	c.452A>C	c.A452C	Y151S
c.416A>T	c.A416T	N139I	c.452A>G	c.A452G	Y151C
c.417T>A	c.T417A	N139K	c.454T>A	c.T454A	Y152N
c.418A>C	c.A418C	K140Q	c.454T>C	c.T454C	Y152H
c.418A>G	c.A418G	K140E	c.454T>G	c.T454G	Y152D
c.419A>C	c.A419C	K140T	c.455A>C	c.A455C	Y152S
c.419A>G	c.A419G	K140R	c.455A>G	c.A455G	Y152C
c.419A>T	c.A419T	K140I	c.455A>T	c.A455T	Y152F
c.420A>T	c.A420T	K140N	c.457G>A	c.G457A	D153N
c.421A>T	c.A421T	T141S	c.457G>C	c.G457C	D153H
c.427G>A	c.G427A	A143T	c.457G>T	c.G457T	D153Y
c.428C>A	c.C428A	A143E	c.458A>C	c.A458C	D153A
c.428C>G	c.C428G	A143G	c.458A>T	c.A458T	D153V
c.428C>T	c.C428T	A143V	c.465T>A または c.465T>G	c.T465A または c.T465G	D155E
c.430G>A	c.G430A	G144S	c.466G>A	c.G466A	A156T
c.430G>C	c.G430C	G144R	c.466G>T	c.G466T	A156S
c.430G>T	c.G430T	G144C	c.467C>G	c.C467G	A156G
c.431G>A	c.G431A	G144D	c.467C>T	c.C467T	A156V
c.431G>C	c.G431C	G144A	c.469C>A	c.C469A	Q157K
c.431G>T	c.G431T	G144V	c.469C>G	c.C469G	Q157E
c.433T>G	c.T433G	F145V	c.470A>C	c.A470C	Q157P
c.434T>A	c.T434A	F145Y	c.470A>T	c.A470T	Q157L

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.471G>C または c.471G>T	c.G471C または c.G471T	Q157H	c.506T>C	c.T506C	F169S
c.472A>G	c.A472G	T158A	c.506T>G	c.T506G	F169C
c.472A>T	c.A472T	T158S	c.507T>A	c.T507A	F169L
c.473C>A	c.C473A	T158N	c.511G>A	c.G511A	G171S
c.473C>T	c.C473T	T158I	c.512G>C	c.G512C	G171A
c.475T>A	c.T475A	F159I	c.512G>T	c.G512T	G171V
c.475T>G	c.T475G	F159V	c.517T>C	c.T517C	Y173H
c.476T>A	c.T476A	F159Y	c.518A>C	c.A518C	Y173S
c.476T>G	c.T476G	F159C	c.518A>G	c.A518G	Y173C
c.477T>A	c.T477A	F159L	c.518A>T	c.A518T	Y173F
c.478G>A	c.G478A	A160T	c.520T>C	c.T520C	C174R
c.478G>T	c.G478T	A160S	c.520T>G	c.T520G	C174G
c.479C>A	c.C479A	A160D	c.523G>C	c.G523C	D175H
c.479C>G	c.C479G	A160G	c.523G>T	c.G523T	D175Y
c.479C>T	c.C479T	A160V	c.524A>G	c.A524G	D175G
c.481G>A	c.G481A	D161N	c.524A>T	c.A524T	D175V
c.481G>C	c.G481C	D161H	c.525C>G または c.525C>A	c.C525G または c.C525A	D175E
c.481G>T	c.G481T	D161Y	c.526A>T	c.A526T	S176C
c.482A>T	c.A482T	D161V	c.528T>A	c.T528A	S176R
c.484T>G	c.T484G	W162G	c.529T>A	c.T529A	L177M
c.485G>C	c.G485C	W162S	c.529T>G	c.T529G	L177V
c.490G>A	c.G490A	V164I	c.530T>C	c.T530C	L177S
c.490G>T	c.G490T	V164L	c.530T>G	c.T530G	L177W
c.491T>C	c.T491C	V164A	c.531G>C	c.G531C	L177F
c.493G>A	c.G493A	D165N	c.532G>A	c.G532A	E178K
c.493G>C	c.G493C	D165H	c.532G>C	c.G532C	E178Q
c.494A>C	c.A494C	D165A	c.533A>C	c.A533C	E178A
c.494A>G	c.A494G	D165G	c.533A>G	c.A533G	E178G
c.495T>A	c.T495A	D165E	c.538T>A	c.T538A	L180M
c.496_497delinsTC	c.496_497delinsTC	L166S	c.538T>G	c.T538G	L180V
c.496C>A	c.C496A	L166M	c.539T>C	c.T539C	L180S
c.496C>G	c.C496G	L166V	c.539T>G	c.T539G	L180W
c.[496C>G; 497T>G]	c.C496G/T497G	L166G	c.540G>C または c.540G>T	c.G540C または c.G540T	L180F
c.497T>A	c.T497A	L166Q	c.541G>A	c.G541A	A181T
c.499C>A	c.C499A	L167I	c.541G>C	c.G541C	A181P
c.499C>G	c.C499G	L167V	c.542C>T	c.C542T	A181V
c.505T>A	c.T505A	F169I	c.544G>T	c.G544T	D182Y
c.505T>G	c.T505G	F169V	c.545A>C	c.A545C	D182A
c.506T>A	c.T506A	F169Y	c.545A>G	c.A545G	D182G



## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.545A>T	c.A545T	D182V	c.571C>A	c.C571A	L191M
c.546T>A	c.T546A	D182E	c.571C>G	c.C571G	L191V
c.548G>A	c.G548A	G183D	c.572T>A	c.T572A	L191Q
c.548G>C	c.G548C	G183A	c.574A>C	c.A574C	N192H
c.550T>A	c.T550A	Y184N	c.574A>G	c.A574G	N192D
c.550T>C	c.T550C	Y184H	c.575A>C	c.A575C	N192T
c.551A>C	c.A551C	Y184S	c.575A>G	c.A575G	N192S
c.551A>G	c.A551G	Y184C	c.576T>A	c.T576A	N192K
c.551A>T	c.A551T	Y184F	c.577A>G	c.A577G	R193G
c.553A>C	c.A553C	K185Q	c.577A>T	c.A577T	R193W
c.553A>G	c.A553G	K185E	c.578G>C	c.G578C	R193T
c.554A>C	c.A554C	K185T	c.578G>T	c.G578T	R193M
c.554A>T	c.A554T	K185M	c.580A>C	c.A580C	T194P
c.555G>C	c.G555C	K185N	c.580A>G	c.A580G	T194A
c.556C>A	c.C556A	H186N	c.580A>T または c.581C>G	c.A580T または c.C581G	T194S
c.556C>G	c.C556G	H186D	c.581C>A	c.C581A	T194N
c.556C>T	c.C556T	H186Y	c.581C>T	c.C581T	T194I
c.557A>T	c.A557T	H186L	c.583G>A	c.G583A	G195S
c.558C>G	c.C558G	H186Q	c.583G>C	c.G583C	G195R
c.559_564dup	c.559_564dup	p.M187_S188dup	c.583G>T	c.G583T	G195C
c.559A>T	c.A559T	M187L	c.584G>T	c.G584T	G195V
c.559A>G	c.A559G	M187V	c.586A>G	c.A586G	R196G
c.560T>C	c.T560C	M187T	c.587G>A	c.G587A	R196K
c.561G>T または c.561G>A または c.561G>C	c.G561T または c.G561A または c.G561C	M187I	c.587G>C	c.G587C	R196T
c.562T>A	c.T562A	S188T	c.587G>T	c.G587T	R196I
c.562T>C	c.T562C	S188P	c.589A>G	c.A589G	S197G
c.562T>G	c.T562G	S188A	c.589A>T	c.A589T	S197C
c.563C>A	c.C563A	S188Y	c.590G>A	c.G590A	S197N
c.563C>G	c.C563G	S188C	c.590G>C	c.G590C	S197T
c.563C>T	c.C563T	S188F	c.590G>T	c.G590T	S197I
c.565T>G	c.T565G	L189V	c.593T>C	c.T593C	I198T
c.566T>C	c.T566C	L189S	c.593T>G	c.T593G	I198S
c.567G>C または c.567G>T	c.G567C または c.G567T	L189F	c.594T>G	c.T594G	I198M
c.568G>A	c.G568A	A190T	c.595G>A	c.G595A	V199M
c.568G>T	c.G568T	A190S	c.595G>C	c.G595C	V199L
c.569C>A	c.C569A	A190D	c.596T>A	c.T596A	V199E
c.569C>G	c.C569G	A190G	c.596T>C	c.T596C	V199A
c.569C>T	c.C569T	A190V	c.596T>G	c.T596G	V199G
			c.598T>A	c.T598A	Y200N

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.599A>C	c.A599C	Y200S	c.632T>G	c.T632G	F211C
c.599A>G	c.A599G	Y200C	c.635A>C	c.A635C	Q212P
c.601T>A	c.T601A	S201T	c.636A>T	c.A636T	Q212H
c.601T>G	c.T601G	S201A	c.637A>C	c.A637C	K213Q
c.602C>A	c.C602A	S201Y	c.637A>G	c.A637G	K213E
c.602C>G	c.C602G	S201C	c.638A>G	c.A638G	K213R
c.602C>T	c.C602T	S201F	c.638A>T	c.A638T	K213M
c.607G>C	c.G607C	E203Q	c.640C>A	c.C640A	P214T
c.608A>C	c.A608C	E203A	c.640C>G	c.C640G	P214A
c.608A>G	c.A608G	E203G	c.640C>T	c.C640T	P214S
c.608A>T	c.A608T	E203V	c.641C>A	c.C641A	P214H
c.609G>C または c.609G>T	c.G609C または c.G609T	E203D	c.641C>G	c.C641G	P214R
c.610T>G	c.T610G	W204G	c.641C>T	c.C641T	P214L
c.611G>C	c.G611C	W204S	c.643A>C	c.A643C	N215H
c.611G>T	c.G611T	W204L	c.643A>G	c.A643G	N215D
c.613C>A	c.C613A	P205T	c.643A>T	c.A643T	N215Y
c.613C>T	c.C613T	P205S	c.644A>C	c.A644C	N215T
c.614C>T	c.C614T	P205L	c.644A>G	c.A644G	N215S
c.616C>A	c.C616A	L206I	c.[644A>G; 937G>T]	c.A644G/G937T	N215S/D313Y*
c.616C>G	c.C616G	L206V	c.644A>T	c.A644T	N215I
c.616C>T	c.C616T	L206F	c.645T>A	c.T645A	N215K
c.617T>A	c.T617A	L206H	c.646T>A	c.T646A	Y216N
c.617T>G	c.T617G	L206R	c.646T>C	c.T646C	Y216H
c.619T>C	c.T619C	Y207H	c.646T>G	c.T646G	Y216D
c.620A>C	c.A620C	Y207S	c.647A>C	c.A647C	Y216S
c.620A>T	c.A620T	Y207F	c.647A>G	c.A647G	Y216C
c.623T>A	c.T623A	M208K	c.647A>T	c.A647T	Y216F
c.623T>G	c.T623G	M208R	c.649A>C	c.A649C	T217P
c.625T>A	c.T625A	W209R	c.649A>G	c.A649G	T217A
c.625T>G	c.T625G	W209G	c.649A>T	c.A649T	T217S
c.627G>C	c.G627C	W209C	c.650C>A	c.C650A	T217K
c.628C>A	c.C628A	P210T	c.650C>G	c.C650G	T217R
c.628C>T	c.C628T	P210S	c.650C>T	c.C650T	T217I
c.629C>A	c.C629A	P210H	c.652G>A	c.G652A	E218K
c.629C>T	c.C629T	P210L	c.652G>C	c.G652C	E218Q
c.631T>C	c.T631C	F211L	c.653A>C	c.A653C	E218A
c.631T>G	c.T631G	F211V	c.653A>G	c.A653G	E218G
c.632T>A	c.T632A	F211Y	c.653A>T	c.A653T	E218V
c.632T>C	c.T632C	F211S	c.654A>T	c.A654T	E218D

\*同一染色体上にある多重変異を表しています。

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.655A>C	c.A655C	I219L	c.694A>G	c.A694G	I232V
c.655A>T	c.A655T	I219F	c.695T>C	c.T695C	I232T
c.656T>A	c.T656A	I219N	c.696T>G	c.T696G	I232M
c.656T>C	c.T656C	I219T	c.698A>C	c.A698C	D233A
c.656T>G	c.T656G	I219S	c.698A>G	c.A698G	D233G
c.657C>G	c.C657G	I219M	c.698A>T	c.A698T	D233V
c.659G>A	c.G659A	R220Q	c.699T>A	c.T699A	D233E
c.659G>C	c.G659C	R220P	c.703T>A	c.T703A	S235T
c.659G>T	c.G659T	R220L	c.703T>G	c.T703G	S235A
c.661C>A	c.C661A	Q221K	c.710A>T	c.A710T	K237I
c.661C>G	c.C661G	Q221E	c.712A>G	c.A712G	S238G
c.662A>C	c.A662C	Q221P	c.712A>T	c.A712T	S238C
c.662A>G	c.A662G	Q221R	c.713G>A	c.G713A	S238N
c.662A>T	c.A662T	Q221L	c.713G>C	c.G713C	S238T
c.663G>C	c.G663C	Q221H	c.713G>T	c.G713T	S238I
c.664T>A	c.T664A	Y222N	c.715A>T	c.A715T	I239L
c.664T>C	c.T664C	Y222H	c.716T>C	c.T716C	I239T
c.664T>G	c.T664G	Y222D	c.717A>G	c.A717G	I239M
c.665A>C	c.A665C	Y222S	c.718A>G	c.A718G	K240E
c.665A>G	c.A665G	Y222C	c.719A>G	c.A719G	K240R
c.670A>C	c.A670C	N224H	c.719A>T	c.A719T	K240M
c.671A>C	c.A671C	N224T	c.720G>C または c.720G>T	c.G720C または c.G720T	K240N
c.671A>G	c.A671G	N224S	c.721A>T	c.A721T	S241C
c.673C>G	c.C673G	H225D	c.722G>C	c.G722C	S241T
c.679C>G	c.C679G	R227G	c.722G>T	c.G722T	S241I
c.682A>C	c.A682C	N228H	c.724A>C	c.A724C	I242L
c.682A>G	c.A682G	N228D	c.724A>G	c.A724G	I242V
c.683A>C	c.A683C	N228T	c.724A>T	c.A724T	I242F
c.683A>G	c.A683G	N228S	c.725T>A	c.T725A	I242N
c.683A>T	c.A683T	N228I	c.725T>C	c.T725C	I242T
c.685T>A	c.T685A	F229I	c.725T>G	c.T725G	I242S
c.686T>A	c.T686A	F229Y	c.726C>G	c.C726G	I242M
c.686T>C	c.T686C	F229S	c.727T>A	c.T727A	L243M
c.687T>A または c.687T>G	c.T687A または c.T687G	F229L	c.727T>G	c.T727G	L243V
c.688G>C	c.G688C	A230P	c.728T>C	c.T728C	L243S
c.689C>A	c.C689A	A230D	c.728T>G	c.T728G	L243W
c.689C>G	c.C689G	A230G	c.729G>C または c.729G>T	c.G729C または c.G729T	L243F
c.689C>T	c.C689T	A230V	c.730G>A	c.G730A	D244N
c.694A>C	c.A694C	I232L	c.730G>C	c.G730C	D244H

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.730G>T	c.G730T	D244Y	c.757A>T	c.A757T	I253F
c.731A>C	c.A731C	D244A	c.758T>A	c.T758A	I253N
c.731A>G	c.A731G	D244G	c.758T>C	c.T758C	I253T
c.731A>T	c.A731T	D244V	c.758T>G	c.T758G	I253S
c.732C>G	c.C732G	D244E	c.760_762delGTT または c.761_763del	c.760_762delGTT または c.761_763del	p.V254del
c.733T>G	c.T733G	W245G	c.760G>T	c.G760T	V254F
c.735G>C	c.G735C	W245C	c.761T>A	c.T761A	V254D
c.736A>G	c.A736G	T246A	c.761T>C	c.T761C	V254A
c.737C>A	c.C737A	T246K	c.761T>G	c.T761G	V254G
c.737C>G	c.C737G	T246R	c.763G>A	c.G763A	D255N
c.737C>T	c.C737T	T246I	c.763G>C	c.G763C	D255H
c.739T>A	c.T739A	S247T	c.763G>T	c.G763T	D255Y
c.739T>G	c.T739G	S247A	c.764A>C	c.A764C	D255A
c.740C>A	c.C740A	S247Y	c.764A>T	c.A764T	D255V
c.740C>G	c.C740G	S247C	c.765T>A	c.T765A	D255E
c.740C>T	c.C740T	S247F	c.766G>C	c.G766C	V256L
c.742T>G	c.T742G	F248V	c.767T>A	c.T767A	V256D
c.743T>A	c.T743A	F248Y	c.767T>G	c.T767G	V256G
c.743T>G	c.T743G	F248C	c.769G>A	c.G769A	A257T
c.744T>A	c.T744A	F248L	c.769G>C	c.G769C	A257P
c.745A>C	c.A745C	N249H	c.769G>T	c.G769T	A257S
c.745A>G	c.A745G	N249D	c.770C>G	c.C770G	A257G
c.745A>T	c.A745T	N249Y	c.770C>T	c.C770T	A257V
c.746A>C	c.A746C	N249T	c.772G>C または c.772G>A	c.G772C または c.G772A	G258R
c.746A>G	c.A746G	N249S	c.773G>A	c.G773A	G258E
c.746A>T	c.A746T	N249I	c.773G>T	c.G773T	G258V
c.747C>G または c.747C>A	c.C747G または c.C747A	N249K	c.775C>A	c.C775A	P259T
c.748C>A	c.C748A	Q250K	c.775C>G	c.C775G	P259A
c.748C>G	c.C748G	Q250E	c.775C>T	c.C775T	P259S
c.749A>C	c.A749C	Q250P	c.776C>A	c.C776A	P259Q
c.749A>G	c.A749G	Q250R	c.776C>G	c.C776G	P259R
c.749A>T	c.A749T	Q250L	c.776C>T	c.C776T	P259L
c.750G>C	c.G750C	Q250H	c.778G>T	c.G778T	G260W
c.751G>A	c.G751A	E251K	c.779G>A	c.G779A	G260E
c.751G>C	c.G751C	E251Q	c.779G>C	c.G779C	G260A
c.752A>G	c.A752G	E251G	c.781G>A	c.G781A	G261S
c.752A>T	c.A752T	E251V	c.781G>C	c.G781C	G261R
c.754A>G	c.A754G	R252G	c.781G>T	c.G781T	G261C
c.757A>G	c.A757G	I253V			

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.782G>C	c.G782C	G261A	c.838C>A	c.C838A	Q280K
c.787A>C	c.A787C	N263H	c.839A>G	c.A839G	Q280R
c.788A>C	c.A788C	N263T	c.839A>T	c.A839T	Q280L
c.788A>G	c.A788G	N263S	c.840A>T または c.840A>C	c.A840T または c.A840C	Q280H
c.790G>A	c.G790A	D264N	c.841G>C	c.G841C	V281L
c.790G>C	c.G790C	D264H	c.842T>A	c.T842A	V281E
c.790G>T	c.G790T	D264Y	c.842T>C	c.T842C	V281A
c.793C>G	c.C793G	P265A	c.842T>G	c.T842G	V281G
c.794C>A	c.C794A	P265Q	c.844A>G	c.A844G	T282A
c.794C>T	c.C794T	P265L	c.844A>T	c.A844T	T282S
c.799A>G	c.A799G	M267V	c.845C>T	c.C845T	T282I
c.799A>T	c.A799T	M267L	c.847C>G	c.C847G	Q283E
c.800T>C	c.T800C	M267T	c.848A>T	c.A848T	Q283L
c.802T>A	c.T802A	L268I	c.849G>C	c.G849C	Q283H
c.804A>T	c.A804T	L268F	c.850A>G	c.A850G	M284V
c.805G>A	c.G805A	V269M	c.850A>T	c.A850T	M284L
c.805G>C	c.G805C	V269L	c.851T>C	c.T851C	M284T
c.806T>C	c.T806C	V269A	c.852G>C	c.G852C	M284I
c.808A>C	c.A808C	I270L	c.853G>A	c.G853A	A285T
c.808A>G	c.A808G	I270V	c.854C>G	c.C854G	A285G
c.809T>C	c.T809C	I270T	c.854C>T	c.C854T	A285V
c.809T>G	c.T809G	I270S	c.856C>G	c.C856G	L286V
c.810T>G	c.T810G	I270M	c.856C>T	c.C856T	L286F
c.811G>A	c.G811A	G271S	c.857T>A	c.T857A	L286H
c.[811G>A; 937G>T]	c.G811A/G937T	G271S/D313Y*	c.860G>T	c.G860T	W287L
c.812G>A	c.G812A	G271D	c.862G>C	c.G862C	A288P
c.812G>C	c.G812C	G271A	c.862G>T	c.G862T	A288S
c.814A>G	c.A814G	N272D	c.863C>G	c.C863G	A288G
c.818T>A	c.T818A	F273Y	c.863C>T	c.C863T	A288V
c.823C>A	c.C823A	L275I	c.865A>C	c.A865C	I289L
c.823C>G	c.C823G	L275V	c.865A>G	c.A865G	I289V
c.827G>A	c.G827A	S276N	c.866T>C	c.T866C	I289T
c.827G>C	c.G827C	S276T	c.866T>G	c.T866G	I289S
c.829T>G	c.T829G	W277G	c.868A>C または c.868A>T	c.A868C または c.A868T	M290L
c.830G>T	c.G830T	W277L	c.868A>G	c.A868G	M290V
c.831G>T または c.831G>C	c.G831T または c.G831C	W277C	c.869T>C	c.T869C	M290T
c.832A>T	c.A832T	N278Y	c.870G>A または c.870G>C または c.870G>T	c.G870A または c.G870C または c.G870T	M290I
c.833A>T	c.A833T	N278I	c.871G>A	c.G871A	A291T
c.835C>G	c.C835G	Q279E			

\*同一染色体上にある多重変異を表しています。

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.871G>T	c.G871T	A291S	c.907A>G	c.A907G	I303V
c.872C>G	c.C872G	A291G	c.907A>T	c.A907T	I303F
c.874G>T	c.G874T	A292S	c.908T>A	c.T908A	I303N
c.875C>G	c.C875G	A292G	c.908T>C	c.T908C	I303T
c.877C>A	c.C877A	P293T	c.908T>G	c.T908G	I303S
c.880T>A	c.T880A	L294I	c.911G>A	c.G911A	S304N
c.880T>G	c.T880G	L294V	c.911G>C	c.G911C	S304T
c.881T>C	c.T881C	L294S	c.911G>T	c.G911T	S304I
c.882A>T	c.A882T	L294F	c.916C>G	c.C916G	Q306E
c.883T>A	c.T883A	F295I	c.917A>C	c.A917C	Q306P
c.883T>G	c.T883G	F295V	c.917A>T	c.A917T	Q306L
c.884T>A	c.T884A	F295Y	c.919G>A	c.G919A	A307T
c.884T>C	c.T884C	F295S	c.919G>C	c.G919C	A307P
c.884T>G	c.T884G	F295C	c.919G>T	c.G919T	A307S
c.886A>G	c.A886G	M296V	c.920C>A	c.C920A	A307D
c.886A>T または c.886A>C	c.A886T または c.A886C	M296L	c.920C>G	c.C920G	A307G
c.887T>C	c.T887C	M296T	c.920C>T	c.C920T	A307V
c.888G>A または c.888G>T または c.888G>C	c.G888A または c.G888T または c.G888C	M296I	c.922A>C	c.A922C	K308Q
c.889T>A	c.T889A	S297T	c.922A>G	c.A922G	K308E
c.892A>G	c.A892G	N298D	c.923A>G	c.A923G	K308R
c.893A>C	c.A893C	N298T	c.923A>T	c.A923T	K308I
c.893A>G	c.A893G	N298S	c.924A>T または c.924A>C	c.A924T または c.A924C	K308N
c.893A>T	c.A893T	N298I	c.925G>A	c.G925A	A309T
c.895G>A	c.G895A	D299N	c.925G>C	c.G925C	A309P
c.895G>C	c.G895C	D299H	c.926C>A	c.C926A	A309D
c.897C>G または c.897C>A	c.C897G または c.C897A	D299E	c.926C>T	c.C926T	A309V
c.898C>A	c.C898A	L300I	c.928C>A	c.C928A	L310I
c.898C>G	c.C898G	L300V	c.928C>G	c.C928G	L310V
c.898C>T	c.C898T	L300F	c.928C>T	c.C928T	L310F
c.899T>C	c.T899C	L300P	c.931C>A	c.C931A	L311I
c.901C>G	c.C901G	R301G	c.931C>G	c.C931G	L311V
c.902G>A	c.G902A	R301Q	c.934C>A	c.C934A	Q312K
c.902G>C	c.G902C	R301P	c.934C>G	c.C934G	Q312E
c.902G>T	c.G902T	R301L	c.935A>G	c.A935G	Q312R
c.904C>A	c.C904A	H302N	c.935A>T	c.A935T	Q312L
c.904C>G	c.C904G	H302D	c.936G>T または c.936G>C	c.G936T または c.G936C	Q312H
c.904C>T	c.C904T	H302Y	c.937G>T	c.G937T	D313Y
c.905A>T	c.A905T	H302L	c.[937G>T; 1232G>A]	c.G937T/G1232A	D313Y/G411D*
			c.938A>G	c.A938G	D313G

\*同一染色体上にある多重変異を表しています。

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化
c.938A>T	c.A938T	D313V
c.939T>A	c.T939A	D313E
c.940A>G	c.A940G	K314E
c.941A>C	c.A941C	K314T
c.941A>T	c.A941T	K314M
c.942G>C	c.G942C	K314N
c.943G>A	c.G943A	D315N
c.943G>C	c.G943C	D315H
c.943G>T	c.G943T	D315Y
c.944A>C	c.A944C	D315A
c.944A>G	c.A944G	D315G
c.944A>T	c.A944T	D315V
c.946G>A	c.G946A	V316I
c.946G>C	c.G946C	V316L
c.947T>C	c.T947C	V316A
c.947T>G	c.T947G	V316G
c.949A>C	c.A949C	I317L
c.949A>G	c.A949G	I317V
c.950T>C	c.T950C	I317T
c.951T>G	c.T951G	I317M
c.952G>A	c.G952A	A318T
c.952G>C	c.G952C	A318P
c.953C>A	c.C953A	A318D
c.953C>T	c.C953T	A318V
c.955A>T	c.A955T	I319F
c.956T>C	c.T956C	I319T
c.957C>G	c.C957G	I319M
c.958A>C	c.A958C	N320H
c.959A>C	c.A959C	N320T
c.959A>G	c.A959G	N320S
c.959A>T	c.A959T	N320I
c.961C>A	c.C961A	Q321K
c.962A>G	c.A962G	Q321R
c.962A>T	c.A962T	Q321L
c.963G>C または c.963G>T	c.G963C または c.G963T	Q321H
c.964G>A	c.G964A	D322N
c.964G>C	c.G964C	D322H
c.965A>C	c.A965C	D322A
c.965A>T	c.A965T	D322V

塩基の変化	塩基の変化	アミノ酸の変化
c.966C>A または c.966C>G	c.C966A または c.C966G	D322E
c.967C>A	c.C967A	P323T
c.968C>G	c.C968G	P323R
c.970T>G	c.T970G	L324V
c.971T>G	c.T971G	L324W
c.973G>A	c.G973A	G325S
c.973G>C	c.G973C	G325R
c.973G>T	c.G973T	G325C
c.974G>C	c.G974C	G325A
c.974G>T	c.G974T	G325V
c.976A>C	c.A976C	K326Q
c.976A>G	c.A976G	K326E
c.977A>C	c.A977C	K326T
c.977A>G	c.A977G	K326R
c.977A>T	c.A977T	K326M
c.978G>C または c.978G>T	c.G978C または c.G978T	K326N
c.979C>G	c.C979G	Q327E
c.980A>C	c.A980C	Q327P
c.980A>T	c.A980T	Q327L
c.981A>T	c.A981T	Q327H
c.983G>C	c.G983C	G328A
c.985T>A	c.T985A	Y329N
c.985T>C	c.T985C	Y329H
c.985T>G	c.T985G	Y329D
c.986A>G	c.A986G	Y329C
c.986A>T	c.A986T	Y329F
c.988C>A	c.C988A	Q330K
c.988C>G	c.C988G	Q330E
c.989A>C	c.A989C	Q330P
c.989A>G	c.A989G	Q330R
c.990G>C	c.G990C	Q330H
c.991C>G	c.C991G	L331V
c.992T>A	c.T992A	L331H
c.992T>C	c.T992C	L331P
c.992T>G	c.T992G	L331R
c.994A>G	c.A994G	R332G
c.995G>C	c.G995C	R332T
c.995G>T	c.G995T	R332I
c.996A>T	c.A996T	R332S

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.997C>G	c.C997G	Q333E	c.1036G>T	c.G1036T	G346C
c.998A>C	c.A998C	Q333P	c.1037G>A	c.G1037A	G346D
c.998A>T	c.A998T	Q333L	c.1037G>C	c.G1037C	G346A
c.1000G>C	c.G1000C	G334R	c.1037G>T	c.G1037T	G346V
c.1001G>A	c.G1001A	G334E	c.1039T>A	c.T1039A	L347I
c.1001G>T	c.G1001T	G334V	c.1043C>A	c.C1043A	A348D
c.1003G>T	c.G1003T	D335Y	c.1046G>C	c.G1046C	W349S
c.1004A>C	c.A1004C	D335A	c.1046G>T	c.G1046T	W349L
c.1004A>G	c.A1004G	D335G	c.1047G>C	c.G1047C	W349C
c.1004A>T	c.A1004T	D335V	c.1048G>A	c.G1048A	A350T
c.1005C>G	c.C1005G	D335E	c.1048G>T	c.G1048T	A350S
c.1006A>G	c.A1006G	N336D	c.1049C>G	c.C1049G	A350G
c.1006A>T	c.A1006T	N336Y	c.1049C>T	c.C1049T	A350V
c.1007A>C	c.A1007C	N336T	c.1052T>A	c.T1052A	V351E
c.1007A>G	c.A1007G	N336S	c.1052T>C	c.T1052C	V351A
c.1007A>T	c.A1007T	N336I	c.1054G>A	c.G1054A	A352T
c.1009T>G	c.T1009G	F337V	c.1054G>T	c.G1054T	A352S
c.1010T>A	c.T1010A	F337Y	c.1055C>G	c.C1055G	A352G
c.1010T>C	c.T1010C	F337S	c.1055C>T	c.C1055T	A352V
c.1010T>G	c.T1010G	F337C	c.1057A>T	c.A1057T	M353L
c.1011T>A	c.T1011A	F337L	c.1058T>A	c.T1058A	M353K
c.1012G>A	c.G1012A	E338K	c.1058T>C	c.T1058C	M353T
c.1013A>C	c.A1013C	E338A	c.1061T>A	c.T1061A	I354K
c.1013A>G	c.A1013G	E338G	c.1061T>G	c.T1061G	I354R
c.1013A>T	c.A1013T	E338V	c.1063A>C	c.A1063C	N355H
c.1014A>T	c.A1014T	E338D	c.1063A>G	c.A1063G	N355D
c.1015G>A	c.G1015A	V339M	c.1063A>T	c.A1063T	N355Y
c.1016T>A	c.T1016A	V339E	c.1064A>G	c.A1064G	N355S
c.1016T>C	c.T1016C	V339A	c.1066C>G	c.C1066G	R356G
c.1021G>C	c.G1021C	E341Q	c.1066C>T	c.C1066T	R356W
c.1022A>C	c.A1022C	E341A	c.1067G>A	c.G1067A	R356Q
c.1027C>A	c.C1027A	P343T	c.1067G>C	c.G1067C	R356P
c.1027C>G	c.C1027G	P343A	c.1067G>T	c.G1067T	R356L
c.1027C>T	c.C1027T	P343S	c.1069C>G	c.C1069G	Q357E
c.1028C>T	c.C1028T	P343L	c.1072G>C	c.G1072C	E358Q
c.1030C>G	c.C1030G	L344V	c.1073A>C	c.A1073C	E358A
c.1030C>T	c.C1030T	L344F	c.1073A>G	c.A1073G	E358G
c.1031T>G	c.T1031G	L344R	c.1074G>T または c.1074G>C	c.G1074T または c.G1074C	E358D
c.1033T>C	c.T1033C	S345P	c.1075A>C	c.A1075C	I359L



## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.1075A>G	c.A1075G	I359V	c.1105G>A	c.G1105A	V369I
c.1075A>T	c.A1075T	I359F	c.1105G>C	c.G1105C	V369L
c.1076T>A	c.T1076A	I359N	c.1105G>T	c.G1105T	V369F
c.1076T>C	c.T1076C	I359T	c.1106T>C	c.T1106C	V369A
c.1076T>G	c.T1076G	I359S	c.1106T>G	c.T1106G	V369G
c.1078G>A	c.G1078A	G360S	c.1108G>A	c.G1108A	A370T
c.1078G>C	c.G1078C	G360R	c.1108G>C	c.G1108C	A370P
c.1078G>T	c.G1078T	G360C	c.1109C>A	c.C1109A	A370D
c.1079G>A	c.G1079A	G360D	c.1109C>G	c.C1109G	A370G
c.1079G>C	c.G1079C	G360A	c.1109C>T	c.C1109T	A370V
c.1082G>A	c.G1082A	G361E	c.1111T>A	c.T1111A	S371T
c.1082G>C	c.G1082C	G361A	c.1112C>G	c.C1112G	S371C
c.1084C>A	c.C1084A	P362T	c.1117G>A	c.G1117A	G373S
c.1084C>G	c.C1084G	P362A	c.1117G>T	c.G1117T	G373C
c.1084C>T	c.C1084T	P362S	c.1118G>C	c.G1118C	G373A
c.1085C>A	c.C1085A	P362H	c.1120A>G	c.A1120G	K374E
c.1085C>G	c.C1085G	P362R	c.1121A>C	c.A1121C	K374T
c.1085C>T	c.C1085T	P362L	c.1121A>G	c.A1121G	K374R
c.1087C>A	c.C1087A	R363S	c.1121A>T	c.A1121T	K374I
c.1087C>G	c.C1087G	R363G	c.1123G>C	c.G1123C	G375R
c.1087C>T	c.C1087T	R363C	c.1124G>A	c.G1124A	G375E
c.1088G>A	c.G1088A	R363H	c.1124G>C	c.G1124C	G375A
c.1088G>T	c.G1088T	R363L	c.1126G>A	c.G1126A	V376M
c.1090T>C	c.T1090C	S364P	c.1126G>C	c.G1126C	V376L
c.1091C>G	c.C1091G	S364C	c.1127T>A	c.T1127A	V376E
c.1093T>A	c.T1093A	Y365N	c.1127T>G	c.T1127G	V376G
c.1093T>G	c.T1093G	Y365D	c.1129G>A	c.G1129A	A377T
c.1094A>C	c.A1094C	Y365S	c.1129G>C	c.G1129C	A377P
c.1094A>T	c.A1094T	Y365F	c.1129G>T	c.G1129T	A377S
c.1096A>C	c.A1096C	T366P	c.1130C>G	c.C1130G	A377G
c.1096A>T	c.A1096T	T366S	c.1135A>G	c.A1135G	N379D
c.1097C>A	c.C1097A	T366N	c.1136A>C	c.A1136C	N379T
c.1097C>T	c.C1097T	T366I	c.1136A>T	c.A1136T	N379I
c.1099A>C	c.A1099C	I367L	c.1137T>A	c.T1137A	N379K
c.1099A>T	c.A1099T	I367F	c.1138C>A	c.C1138A	P380T
c.1101C>G	c.C1101G	I367M	c.1138C>G	c.C1138G	P380A
c.1102G>A	c.G1102A	A368T	c.1139C>A	c.C1139A	P380H
c.1102G>C	c.G1102C	A368P	c.1139C>G	c.C1139G	P380R
c.1103C>G	c.C1103G	A368G	c.1139C>T	c.C1139T	P380L

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化	塩基の変化	塩基の変化	アミノ酸の変化
c.1142C>A	c.C1142A	A381D	c.1181T>C	c.T1181C	L394P
c.1147T>A	c.T1147A	F383I	c.1181T>G	c.T1181G	L394R
c.1148T>A	c.T1148A	F383Y	c.1183G>C	c.G1183C	G395R
c.1148T>G	c.T1148G	F383C	c.1184G>A	c.G1184A	G395E
c.1150A>T	c.A1150T	I384F	c.1184G>C	c.G1184C	G395A
c.1151T>C	c.T1151C	I384T	c.1186T>A	c.T1186A	F396I
c.1152C>G	c.C1152G	I384M	c.1186T>G	c.T1186G	F396V
c.1153A>G	c.A1153G	T385A	c.1187T>G	c.T1187G	F396C
c.1154C>T	c.C1154T	T385I	c.1188C>G	c.C1188G	F396L
c.1156C>A	c.C1156A	Q386K	c.1189T>A	c.T1189A	Y397N
c.1157A>T	c.A1157T	Q386L	c.1189T>C	c.T1189C	Y397H
c.1158G>C	c.G1158C	Q386H	c.1190A>C	c.A1190C	Y397S
c.1159C>A	c.C1159A	L387I	c.1190A>G	c.A1190G	Y397C
c.1159C>T	c.C1159T	L387F	c.1190A>T	c.A1190T	Y397F
c.1160T>A	c.T1160A	L387H	c.1192G>A	c.G1192A	E398K
c.1160T>G	c.T1160G	L387R	c.1192G>C	c.G1192C	E398Q
c.1162C>A	c.C1162A	L388I	c.1193A>G	c.A1193G	E398G
c.1162C>G	c.C1162G	L388V	c.1195T>A	c.T1195A	W399R
c.1162C>T	c.C1162T	L388F	c.1195T>G	c.T1195G	W399G
c.1163T>A	c.T1163A	L388H	c.1198A>C	c.A1198C	T400P
c.1163T>G	c.T1163G	L388R	c.1198A>G	c.A1198G	T400A
c.1168G>A	c.G1168A	V390M	c.1198A>T	c.A1198T	T400S
c.1171A>C	c.A1171C	K391Q	c.1199C>A	c.C1199A	T400N
c.1171A>G	c.A1171G	K391E	c.1199C>T	c.C1199T	T400I
c.1172A>C	c.A1172C	K391T	c.1201T>A	c.T1201A	S401T
c.1172A>G	c.A1172G	K391R	c.1201T>G	c.T1201G	S401A
c.1172A>T	c.A1172T	K391I	c.1202_1203insGACTTC	c.1202_1203insGACTTC	p.T400_S401dup
c.1173A>T	c.A1173T	K391N	c.1202C>T	c.C1202T	S401L
c.1174A>G	c.A1174G	R392G	c.1204A>G	c.A1204G	R402G
c.1174A>T	c.A1174T	R392W	c.1204A>T	c.A1204T	R402W
c.1175G>A	c.G1175A	R392K	c.1205G>C	c.G1205C	R402T
c.1175G>C	c.G1175C	R392T	c.1205G>T	c.G1205T	R402M
c.1175G>T	c.G1175T	R392M	c.1206G>C	c.G1206C	R402S
c.1177A>C	c.A1177C	K393Q	c.1207T>G	c.T1207G	L403V
c.1177A>G	c.A1177G	K393E	c.1208T>C	c.T1208C	L403S
c.1178A>C	c.A1178C	K393T	c.1209A>T	c.A1209T	L403F
c.1179G>C	c.G1179C	K393N	c.1210A>G	c.A1210G	R404G
c.1180C>A	c.C1180A	L394I	c.1211G>A	c.C1211A	R404K
c.1181T>A	c.T1181A	L394Q	c.1211G>C	c.C1211C	R404T

## ガラフォルド®に反応性のあるGLA変異一覧

塩基の変化	塩基の変化	アミノ酸の変化
c.1211G>T	c.G1211T	R404I
c.1212A>T	c.A1212T	R404S
c.1213A>G	c.A1213G	S405G
c.1216C>G	c.C1216G	H406D
c.1217A>T	c.A1217T	H406L
c.1218C>G	c.C1218G	H406Q
c.1219A>T	c.A1219T	I407L
c.1220T>C	c.T1220C	I407T
c.1221A>G	c.A1221G	I407M
c.1222A>C	c.A1222C	N408H
c.1222A>G	c.A1222G	N408D
c.1222A>T	c.A1222T	N408Y
c.1223A>C	c.A1223C	N408T
c.1225C>A	c.C1225A	P409T
c.1225C>G	c.C1225G	P409A
c.1225C>T	c.C1225T	P409S
c.1226C>T	c.C1226T	P409L
c.1228A>G	c.A1228G	T410A
c.1228A>T	c.A1228T	T410S
c.1229C>T	c.C1229T	T410I
c.1231G>A	c.G1231A	G411S
c.1231G>T	c.G1231T	G411C
c.1232G>A	c.G1232A	G411D
c.1232G>C	c.G1232C	G411A
c.1232G>T	c.G1232T	G411V
c.1234A>C	c.A1234C	T412P
c.1234A>G	c.A1234G	T412A
c.1234A>T	c.A1234T	T412S
c.1235C>A	c.C1235A	T412N
c.1235C>T	c.C1235T	T412I
c.1237G>A	c.G1237A	V413I
c.1237G>T	c.G1237T	V413F
c.1238T>G	c.T1238G	V413G
c.1240T>G	c.T1240G	L414V
c.1242G>C	c.G1242C	L414F
c.1243C>A	c.C1243A	L415I
c.1244T>A	c.T1244A	L415H
c.1246C>G	c.C1246G	Q416E
c.1247A>T	c.A1247T	Q416L

塩基の変化	塩基の変化	アミノ酸の変化
c.1248G>C	c.G1248C	Q416H
c.1249C>A	c.C1249A	L417I
c.1252G>A	c.G1252A	E418K
c.1252G>C	c.G1252C	E418Q
c.1253A>C	c.A1253C	E418A
c.1253A>G	c.A1253G	E418G
c.1254A>T	c.A1254T	E418D
c.1255A>G	c.A1255G	N419D
c.1255A>T	c.A1255T	N419Y
c.1256A>C	c.A1256C	N419T
c.1256A>G	c.A1256G	N419S
c.1256A>T	c.A1256T	N419I
c.1258A>C	c.A1258C	T420P
c.1258A>T	c.A1258T	T420S
c.1259C>A	c.C1259A	T420K
c.1259C>G	c.C1259G	T420R
c.1261A>G	c.A1261G	M421V
c.1261A>T	c.A1261T	M421L
c.1262T>A	c.T1262A	M421K
c.1262T>C	c.T1262C	M421T
c.1262T>G	c.T1262G	M421R
c.1263G>C	c.G1263C	M421I
c.1265A>C	c.A1265C	Q422P
c.1267A>T	c.A1267T	M423L
c.1268T>A	c.T1268A	M423K
c.1268T>C	c.T1268C	M423T
c.1269G>C	c.G1269C	M423I
c.1271C>T	c.C1271T	S424L
c.1275A>C	c.A1275C	L425F
c.1279G>A	c.G1279A	D427N
c.1286T>G	c.T1286G	L429R

ファブリー病治療剤 処方箋医薬品<sup>※</sup>ガラフォルド<sup>®</sup> カプセル123mgGALAFOLD<sup>®</sup> Capsules 123mg

ミゲラスタット塩酸塩

日本標準商品分類番号	873999
承認番号	23000AMX00463000
薬価収載	2018年5月
販売開始	2018年5月

貯法：室温保存  
有効期間：4年  
注) 注意-医師等の処方箋により使用すること

## 2. 禁忌(次の患者には投与しないこと)


本剤の成分に対し過敏症の既往歴のある患者

## 3. 組成・性状

## 3.1 組成

有効成分 (1カプセル中)	ミゲラスタット塩酸塩150mg (ミゲラスタットとして123mg)
添加剤	内容物：部分アルファー化デンプン、ステアリン酸マグネシウム カプセル：ゼラチン、酸化チタン、青色二色

## 3.2 製剤の性状

色・剤形	ボディが白色不透明でキャップが青色不透明の硬カプセル
外形	
大きさ	6.4×18.0mm(2号)
識別コード	A1001

## 4. 効能又は効果

ミゲラスタットに反応性のあるGLA遺伝子変異を伴うファブリー病

## 5. 効能又は効果に関連する注意

- 5.1 本剤はファブリー病と確定診断された患者に対して使用すること。  
5.2 本剤の投与開始に先立って、患者のGLA遺伝子変異のミゲラスタットに対する反応性を確認すること。[24.参照]

## \* 6. 用法及び用量

通常、成人及び12歳以上の小児にはミゲラスタットとして1回123mgを隔日経口投与する。なお、食事の前後2時間を避けて投与すること。

## 7. 用法及び用量に関連する注意

- 7.1 本剤の曝露量は食事の影響を受けるため、食事の前後2時間を避けて投与すること。[16.2.1参照]  
7.2 投与時刻は原則毎回一定とする。服用予定時刻に服用できなかった場合は、服用予定時刻から12時間以内に服用すること。服用予定時刻から12時間を超えた場合は、次の服用予定日時から服用を再開すること。  
7.3 酵素補充療法との併用に関する有効性及び安全性は確立されていない。

## 8. 重要な基本的注意

本剤投与中は、定期的に腎機能、心機能、臨床検査値等を確認する等経過を十分に観察し、本剤投与で効果が認められない場合には治療法の変更を考慮すること。

## 9. 特定の背景を有する患者に関する注意

## 9.2 腎機能障害患者

## 9.2.1 重度の腎機能障害患者

重度の腎機能障害のある患者は有効性及び安全性を指標とした臨床試験では除外されている。本剤の血中濃度が上昇するおそれがあることから、重度の腎機能障害のある患者への投与は推奨されない。[16.6.1、17.1.1、17.1.2、18.1、18.2参照]

## 9.5 妊婦

妊婦又は妊娠している可能性のある女性には、治療上の有益性が危険性を上回ると判断される場合のみ使用すること。ウサギ胚胎児発生に関する試験において、臨床推奨用量投与時の曝露量の244倍以上に相当するミゲラスタットの投与により、母動物毒性が発現し、着床後胚死亡率の増加、体重減少、流産、骨化遅延、軽微な骨異常の増加等が認められた。

## 9.6 授乳婦

授乳しないことが望ましい。ラットで乳汁へ移行することが報告されている。

## \* 9.7 小児等

本剤の曝露量は体重の影響を受ける可能性があることから、12歳以上の小児には患者の体重に留意した上で投与の適否を判断し、投与にあたっては患者の状態を十分に観察すること。12歳以上16歳未満の患者では、体重45kg未満の患者は臨床試験に組み入れられていない。また、12歳未満の小児等を対象とした臨床試験は実施していない。[16.1.3、17.1.3参照]

## 9.8 高齢者

患者の状態を観察しながら慎重に投与すること。一般に生理機能が低下していることが多い。

## 10. 相互作用

## \*\* 10.2 併用注意(併用に注意すること)

薬剤名等	臨床症状・措置方法	機序・危険因子
カフェイン [16.7.2参照]	カフェインとの同時摂取により、本剤のC <sub>max</sub> 及びAU <sub>C<sub>0-12h</sub></sub> がそれぞれ60%及び55%減少したとの報告があることから、本剤服用の前後2時間は摂取を避けること。	機序は不明である。

## \* 11. 副作用

次の副作用があらわれることがあるので、観察を十分に行い、異常が認められた場合には投与を中止するなど適切な処置を行うこと。

## 11.2 その他の副作用

	10%以上	5%以上10%未満	1%以上5%未満
心臓			動悸
耳および迷路			回転性めまい
* 胃腸		下痢	悪心、腹痛、便秘、口内乾燥、便秘切迫、消化不良、嘔吐
一般・全身			疲労、疼痛
* 臨床検査			血中クレアチンホスホキナーゼ増加、体重増加、血圧上昇
筋骨格系および結合組織			筋痙攣、筋肉痛、斜頸、四肢痛
* 神経系	頭痛	錯感覚	浮動性めまい、感覚鈍麻
精神			うつ病
腎および尿路			蛋白尿
呼吸器、胸郭および縦隔			呼吸困難、鼻出血
皮膚および皮下組織			発疹、そう痒症

## 15. その他の注意

## 15.2 非臨床試験に基づく情報

雄ラットの受胎能及び初期胚発生に関する試験において、臨床推奨用量投与時の曝露量の0.163倍、1.5倍に相当するミゲラスタットの投与により、受胎率の低下(それぞれ75%、55%)が認められた。受胎率の低下は投与中止4週間後に回復した。

## 21. 承認条件

- 21.1 医薬品リスク管理計画を策定の上、適切に実施すること。  
21.2 国内での治験症例が極めて限られていることから、製造販売後、再審査期間中の全投与症例を対象に使用成績調査を実施することにより、本剤の使用患者の背景情報を把握するとともに、本剤の安全性及び有効性に関するデータを早期に収集し、本剤の適正使用に必要な措置を講じること。

## 22. 包装

7カプセル 1シート PTP

## 24. 文献請求先及び問い合わせ先

文献請求先：

アマカス・セラピューティクス株式会社

コールセンター

〒100-0005 東京都千代田区丸の内一丁目6番2号

新丸の内センタービルディング19階

フリーダイヤル 0120-907-477

受付時間：9:00～17:00(土・日・祝日及び会社休日を除く)

お問い合わせ先(GLA遺伝子変異のミゲラスタットに対する反応性に関するお問い合わせ)：

患者のGLA遺伝子変異のミゲラスタットに対する反応性は、アマカス・セラピューティクス株式会社が提供する最新の「ガラフォルドに反応性のあるGLA変異一覧表」又は専用のウェブサイト(www.galafoldamenabilitytable.com)を確認してください。「ガラフォルドに反応性のあるGLA変異一覧表」又は専用のウェブサイトには患者のGLA遺伝子変異が未掲載であった場合等には、下記にお問い合わせください。[5.2参照]

コールセンター

フリーダイヤル 0120-907-477 e-mail info@amicusrx.jp

## 26. 製造販売業者等

## 26.1 製造販売元

アマカス・セラピューティクス株式会社

〒100-0005 東京都千代田区丸の内一丁目6番2号

新丸の内センタービルディング19階

\*\* 2023年11月改訂(第3版)

\* 2022年12月改訂(第2版)