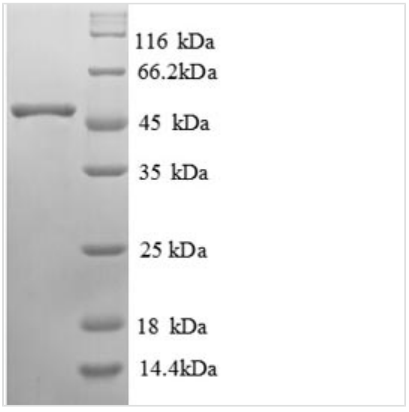
Recombinant Human Alpha-galactosidase A (GLA)

Product Code	CSB-EP009474HU
Abbreviation	Recombinant Human GLA protein
Storage	The shelf life is related to many factors, storage state, buffer ingredients, storage temperature and the stability of the protein itself. Generally, the shelf life of liquid form is 6 months at -20°C/-80°C. The shelf life of lyophilized form is 12 months at -20°C/-80°C.
Uniprot No.	P06280
Product Type	Recombinant Proteins
Immunogen Species	Homo sapiens (Human)
Purity	Greater than 90% as determined by SDS-PAGE.
Sequence	LDNGLARTPTMGWLHWERFMCNLDCQEEPDSCISEKLFMEMAELMVSEGWK DAGYEYLCIDDCWMAPQRDSEGRLQADPQRFPHGIRQLANYVHSKGLKLGIY ADVGNKTCAGFPGSFGYYDIDAQTFADWGVDLLKFDGCYCDSLENLADGYKH MSLALNRTGRSIVYSCEWPLYMWPFQKPNYTEIRQYCNHWRNFADIDDSWKS IKSILDWTSFNQERIVDVAGPGGWNDPDMLVIGNFGLSWNQQVTQMALWAIM AAPLFMSNDLRHISPQAKALLQDKDVIAINQDPLGKQGYQLRQGDNFEVWERP LSGLAWAVAMINRQEIGGPRSYTIAVASLGKGVACNPACFITQLLPVKRKLGFY EWTSRLRSHINPTGTVLLQLENTMQMSLKDLL
Research Area	Cardiovascular
Source	E.coli
Target Names	GLA
Expression Region	32-429aa
Notes	Repeated freezing and thawing is not recommended. Store working aliquots at 4°C for up to one week.
Tag Info	N-terminal 6xHis-tagged
Mol. Weight	49.4kDa
Protein Length	Full Length of Mature Protein
Image	 (Tris-Glycine gel) Discontinuous SDS-PAGE (reduced) with 5% enrichment gel and 15% separation gel.

Description

Alpha-galactosidase A (GLA) is a crucial enzyme involved in lysosomal function and lipid metabolism. GLA is responsible for hydrolyzing the terminal alpha-galactosyl moiety from glycoconjugates [1]. Mutations in the GLA gene lead to Fabry disease, a rare X-linked disorder characterized by deficient activity of GLA, resulting in the intracellular accumulation of enzyme substrates inside lysosomes [2]. This accumulation, particularly of globotriaosylceramide-3 (Gb3), is specific to Fabry disease with classical mutations and is associated with small fiber neuropathy [3].

Fabry disease can present with symptoms mimicking hypertrophic cardiomyopathy, emphasizing the importance of genetic screening to establish a diagnosis, especially in women [4]. The absence or deficiency of GLA enzyme due to mutations results in multiorgan glycosphingolipid accumulations, leading to various clinical manifestations [5]. Chaperone therapy has been explored in Fabry disease to address the underlying enzyme deficiency [6].

The cardiovascular phenotype in Fabry disease has been linked to residual GLA activity, which may influence disease progression and the manifestation of clinical signs [7]. Dysregulation of immune response mediators and pain-related ion channels has been associated with pain-like behavior in Fabry disease, highlighting the complex interplay between GLA mutations and symptomatology [8]. Additionally, cornea verticillata has been reported in classical Fabry disease cases, further underlining the diverse systemic manifestations of GLA mutations.

References:
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associated with pain-like behavior in the gla ko mouse model of fabry disease", Cells, vol. 11, no. 11, p. 1730, 2022. https://doi.org/10.3390/cells11111730
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Shelf Life

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