GCS1 Antibody (N-term)

Catalog_no： AB2065

Reactivity： H

Category： 抗原抗体

Size： 100μL/50μL

Immunogen： HUMAN:1-30

Specificity： This GCS1 antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 1-30 amino acids from the N-terminal region of human GCS1.

Dilution： WB,1:1000;

Purification： Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein G column, eluted with high and low pH buffers and neutralized immediately, followed by dialysis against PBS.

Other_name： Mannosyl-oligosaccharide glucosidase, Processing A-glucosidase I, MOGS, GCS1

Isotype： Rabbit Ig

Background： GCS1 cleaves the distal alpha 1,2-linked glucose residue from the Glc(3)Man(9)GlcNAc(2) oligosaccharide precursor in a highly specific manner. Defects in GCS1 are the cause of type IIb congenital disorder of glycosylation (CDGIIb). This syndrome is also known as glucosidase I deficiency and is characterized by marked generalized hypotonia and hypomotility of the neonate, dysmorphic features, including a prominent occiput, short palpebral fissures, retrognathia, high arched palate, generalized edema, and hypoplastic genitalia. Symptoms include hepatomegaly, hypoventilation, feeding problems and seizures. The clinical course is progressive and survival is at most a few months.

reference： Volker, C., et al., Glycobiology 12(8):473-483 (2002). De Praeter, C.M., et al., Am. J. Hum. Genet. 66(6):1744-1756 (2000). Kalz-Fuller, B., et al., Eur. J. Biochem. 231(2):344-351 (1995). Kalz-Fueller, B., et al., Eur. J. Biochem. 249, 912-912 (1997