B4GALT7 cDNA Clone – Xylosylprotein Beta 1,4-Galactosyltransferase, Polypeptide 7 (Galactosyltransferase I) (Human)

PRODUCT OVERVIEW

Product Name	B4GALT7, cDNA Clone
Full Product Name	B4GALT7 cDNA Clone
Product Gene Name	B4GALT7 cdna clone
Sequence	atgttcccct cgcggaggaa agcggcgcag ctgccctggg aggacggcag gtccgggttg ctctccggcg gcctccctcg gaagtgttcc gtcttccacc tgttcgtggc ctgcctctcg ctgggcttct tctccctact ctggctgcag ctcagctgct ctggggacgt ggcccgggca gtcaggggac aagggcagga gacctcgggc cctccccgcg cctgcccccc agagccgccc cctgagcact gggaagaaga cgcatcctgg ggcccccacc gcctggcagt gctggtgccc ttccgcgaac gcttcgagga gctcctggtc ttcgtgcccc acatgcgccg cttcctgagc aggaagaaga tccggcacca catctacgtg ctcaaccagg tggaccactt caggttcaac cgggcagcgc tcatcaacgt gggcttcctg gagagcagca acagcacgga ctacattgcc atgcacgacg ttgacctgct ccctctcaac gaggagctgg actatggctt tcctgaggct gggcccttcc acgtggcctc cccggagctc caccctctct accactacaa gacctatgtc ggcggcatcc tgctgctctc caagcagcac taccggctgt gcaatgggat gtccaaccgc ttctggggct ggggccgcga ggacgacgag ttctaccggc gcattaaggg agctgggctc cagcttttcc gcccctcggg aatcacaact gggtacaaga catttcgcca cctgcatgac ccagcctggc ggaagaggga ccagaagcgc atcgcagctc aaaaacagga gcagttcaag gtggacaggg agggaggcct gaacactgtg aagtaccatg tggcttcccg cactgccctg tctgtgggcg gggccccctg cactgtcctc aacatcatgt tggactgtga caagaccgcc acaccctggt gcacattcag ctga
Molecular Weight	37,406 Da
OMIM	130070
Vector	pENTR223.1 or pUC
Clone Sequence Report	Provided with product shipment
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Related Product Information for B4GALT7 cdna clone	MBS1265982 is a cDNA clone targeting B4GALT7.
Other Notes	Small volumes of B4GALT7 cdna clone vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for B4GALT7. It may not necessarily be applicable to this product.

NCBI GI #	33873618
NCBI GeneID	11285
NCBI Accession #	BC007317
UniProt Secondary Accession #	Q9UHN2; B3KN39
UniProt Related Accession #	Q9UBV7
NCBI Official Full Name	Homo sapiens xylosylprotein beta 1,4-galactosyltransferase, polypeptide 7 (galactosyltransferase I), mRNA
NCBI Official Synonym Full Names	beta-1,4-galactosyltransferase 7
NCBI Official Symbol	B4GALT7
NCBI Official Synonym Symbols	EDSP1; XGPT1; EDSSLA; XGALT1
UniProt Protein Name	Beta-1,4-galactosyltransferase 7

UniProt Synonym Protein Names	UDP-Gal:beta-GlcNAc beta-1,4-galactosyltransferase 7; UDP-galactose:beta-N-acetylglucosamine beta-1,4-galactosyltransferase 7Including the following 1 domains:Xylosylprotein 4-beta-galactosyltransferase (EC:2.4.1.133)Alternative name(s):Proteoglycan UDP-galactose:beta-xylose beta1,4-galactosyltransferase I; UDP-galactose:beta-xylose beta-1,4-galactosyltransferase; XGPT; XGalT-1; Xylosylprotein beta-1,4-galactosyltransferase
Protein Family	Beta-1,4-galactosyltransferase
UniProt Gene Name	B4GALT7
UniProt Synonym Gene Names	XGALT1; Beta-1,4-GalTase 7; Beta4Gal-T7; b4Gal-T7
UniProt Entry Name	B4GT7_HUMAN
NCBI Protein Information	beta-1,4-galactosyltransferase 7
NCBI Summary for B4GALT7	This gene is a member of the beta-1,4-galactosyltransferase (beta4GalT) family. Family members encode type II membrane-bound glycoproteins that appear to have exclusive specificity for the donor substrate UDP-galactose. Each beta4GalT member has a distinct function in the biosynthesis of different glycoconjugates and saccharide structures. As type II membrane proteins, they have an N-terminal hydrophobic signal sequence that directs the protein to the Golgi apparatus which then remains uncleaved to function as a transmembrane anchor. The enzyme encoded by this gene attaches the first galactose in the common carbohydrate-protein linkage (GlcA-beta1,3-Gal-beta1,3-Gal-beta1,4-Xyl-beta1-O-Ser) found in proteoglycans. This enzyme differs from other beta4GalTs because it lacks the conserved Cys residues found in beta4GalT1-beta4GalT6 and it is located in cis-Golgi instead of trans-Golgi. Mutations in this gene have been associated with the progeroid form of Ehlers-Danlos syndrome. [provided by RefSeq, Oct 2009]
UniProt Comments for B4GALT7	B4GALT7: Required for the biosynthesis of the tetrasaccharide linkage region of proteoglycans, especially for small proteoglycans in skin fibroblasts. Defects in B4GALT7 are the cause of Ehlers-Danlos syndrome progeroid type (EDSP). EDSP is a variant form of Ehlers-Danlos syndrome characterized by progeroid facies, mild mental retardation, short stature, skin hyperextensibility, moderate skin fragility, joint hypermobility principally in digits. Belongs to the glycosyltransferase 7 family. Protein type: Transferase; EC 2.4.1.133; Glycan Metabolism - chondroitin sulfate biosynthesis; Glycan Metabolism - heparan sulfate biosynthesis; Membrane protein, integral Chromosomal Location of Human Ortholog: 5q35.2-q35.3 Cellular Component: Golgi apparatus; Golgi membrane; integral to membrane Molecular Function: beta-N-acetylglucosaminylglycopeptide beta-1,4-galactosyltransferase activity; galactosyltransferase activity; manganese ion binding; protein binding; xylosylprotein 4-beta-galactosyltransferase activity Biological Process: fibril organization and biogenesis; glycosaminoglycan biosynthetic process; glycosaminoglycan metabolic process; negative regulation of fibroblast proliferation; protein amino acid N-linked glycosylation; protein modification process; proteoglycan metabolic process Disease: Ehlers-danlos Syndrome, Progeroid Type, 1
Research Articles on B4GALT7	**1.** Our findings demonstrate that B4GALT7 is the causative gene for LRS. The identification of a unique homozygous mutation argues in favor of a founder effect. B4GALT7 encodes a galactosyltransferase.
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