

Product datasheet

Recombinant Human LRP5 protein ab114399

1 Image

Description	
Product name	Recombinant Human LRP5 protein
Expression system	Wheat germ
Accession	<u><b>O75197</b></u>
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	RDVRLVDAGGVKLESTMVSGLEDAAAVDFQFSKGAVYW TDVSEEAIKQT YLNQTGAAVQNVVISGLVSPDGLACDWVGKKLYWTDSET N
Predicted molecular weight	36 kDa including tags
Amino acids	41 to 130
Tags	GST tag N-Terminus

Specifications	
Our <b><u>Abpromise guarantee</u></b> covers the use of <b>ab114399</b> in the following tested applications.	
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.	
Applications	SDS-PAGE ELISA Western blot
Form	Liquid
Additional notes	Protein concentration is lot specific and is between 0.1 - 0.58 ug/uL

Preparation and Storage	
Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles. pH: 8.00

## General Info

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### Function

Component of the Wnt-Fzd-LRP5-LRP6 complex that triggers beta-catenin signaling through inducing aggregation of receptor-ligand complexes into ribosome-sized signalsomes. Cell-surface coreceptor of Wnt/beta-catenin signaling, which plays a pivotal role in bone formation. The Wnt-induced Fzd/LRP6 coreceptor complex recruits DVL1 polymers to the plasma membrane which, in turn, recruits the AXIN1/GSK3B-complex to the cell surface promoting the formation of signalsomes and inhibiting AXIN1/GSK3-mediated phosphorylation and destruction of beta-catenin. Appears to be required for postnatal control of vascular regression in the eye. Required for posterior patterning of the epiblast during gastrulation.

### Tissue specificity

Widely expressed, with the highest level of expression in the liver.

### Involvement in disease

Defects in LRP5 are the cause of vitreoretinopathy exudative type 4 (EVR4) [MIM:601813]. EVR4 is a disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease-related abnormality is an arc of avascular retina in the extreme temporal periphery. EVR4 inheritance can be autosomal dominant or recessive.

Genetic variations in LRP5 are a cause of susceptibility to osteoporosis (OSTEOP) [MIM:166710]; also known as senile osteoporosis or postmenopausal osteoporosis.

Osteoporosis is characterized by reduced bone mass, disruption of bone microarchitecture without alteration in the composition of bone. Osteoporotic bones are more at risk of fracture.

Defects in LRP5 are the cause of osteoporosis-pseudoglioma syndrome (OPPG) [MIM:259770]; also known as osteogenesis imperfecta ocular form. OPPG is a recessive disorder characterized by very low bone mass and blindness. Individuals with OPPG are prone to develop bone fractures and deformations and have various eye abnormalities, including phthisis bulbi, retinal detachments, falciform folds or persistent vitreal vasculature.

Defects in LRP5 are a cause of high bone mass trait (HBM) [MIM:601884]. HBM is a rare phenotype characterized by exceptionally dense bones. HBM individuals show otherwise a completely normal skeletal structure and no other unusual clinical findings.

Defects in LRP5 are a cause of endosteal hyperostosis Worth type (WENHY) [MIM:144750]; also known as autosomal dominant osteosclerosis. WENHY is an autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity.

Defects in LRP5 are the cause of osteopetrosis autosomal dominant type 1 (OPTA1) [MIM:607634]. Osteopetrosis is a rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTA1 is characterized by generalized osteosclerosis most pronounced in the cranial vault. Patients are often asymptomatic, but some

suffer from pain and hearing loss. It appears to be the only type of osteopetrosis not associated with an increased fracture rate.

Defects in LRP5 are the cause of van Buchem disease type 2 (VBCH2)[MIM:607636]. VBCH2 is an autosomal dominant sclerosing bone dysplasia characterized by cranial osteosclerosis, thickened calvaria and cortices of long bones, enlarged mandible and normal serum alkaline phosphatase levels.

#### Sequence similarities

Belongs to the LDLR family.

Contains 4 EGF-like domains.

Contains 3 LDL-receptor class A domains.

Contains 20 LDL-receptor class B repeats.

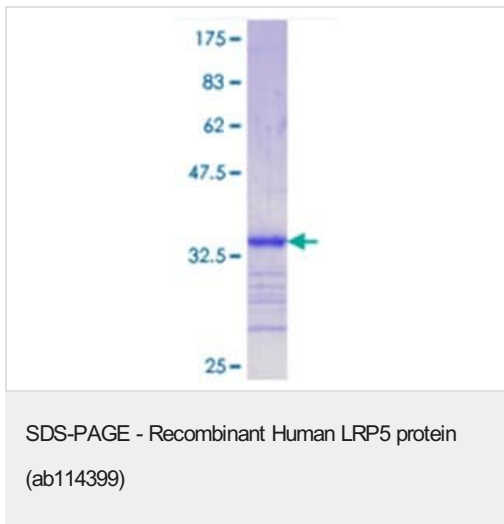
#### Post-translational modifications

Phosphorylation of cytoplasmic PPPSP motifs regulates the signal transduction of the Wnt signaling pathway through acting as a docking site for AXIN1.

#### Cellular localization

Membrane. Endoplasmic reticulum. Chaperoned to the plasma membrane by MESD.

### Images



12.5% SDS-PAGE analysis of ab114399, stained with Coomassie Blue.

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