

Rabbit Anti-SHOX antibody

SL17470R

Product Name:	SHOX
Chinese Name:	X连锁生长调控因子GCFX抗体
Alias:	GCFX; Growth control factor, X linked; Homo sapiens shox gene, alternatively spliced products, complete cds; PHOG; Pseudoautosomal homeobox containing osteogenic; Pseudoautosomal homeobox containing osteogenic protein; Pseudoautosomal homeobox-containing osteogenic protein; Short stature homeobox containing protein; Short stature homeobox protein; Short stature homeobox-containing protein; SHOX; SHOX_HUMAN; SHOXY; SS; Turner syndrome.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Horse,
Applications:	IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	32kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SHOX:1-100/292
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	This gene belongs to the paired homeobox family and is located in the pseudoautosomal region 1 (PAR1) of X and Y chromosomes. Defects in this gene are

associated with idiopathic growth retardation and in the short stature phenotype of Turner syndrome patients. This gene is highly conserved across species from mammals to fish to flies. Alternatively spliced transcript variants encoding different isoforms have been noted for this gene. [provided by RefSeq, Jul 2008]

Function:

Controls fundamental aspects of growth and development.

Subcellular Location:

Nucleus.

Tissue Specificity:

SHOXA is expressed in skeletal muscle, placenta, pancreas, heart and bone marrow fibroblast and SHOXB is highly expressed in bone marrow fibroblast followed by kidney and skeletal muscle. SHOXB is not expressed in brain, kidney, liver and lung. Highly expressed in osteogenic cells.

DISEASE:

Defects in SHOX are the cause of Leri-Weill dyschondrosteosis (LWD) [MIM:127300].

LWD is a dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. Defects in SHOX are a cause of Langer mesomelic dysplasia (LMD) [MIM:249700]. LMD is an autosomal recessive rare skeletal dysplasia characterized by severe short stature owing to shortening and maldevelopment of the mesomelic and rhizomelic segments of the limbs. Associated malformations are rarely reported and intellect is normal in all affected subjects reported to date.

Defects in SHOX are a cause of idiopathic short stature (ISS) [MIM:300582]. Idiopathic short stature is usually defined as a height below the third percentile for chronological age or minus 2 standard deviations of national height standards in the absence of specific causative disorders.

Similarity:

Belongs to the paired homeobox family. Bicoid subfamily. Contains 1 homeobox DNA-binding domain.

SWISS:

O15266

Gene ID: 6473

Database links:

Entrez Gene: 615159 Cow
Entrez Gene: 491706 Dog
Entrez Gene: 6473 Human
<u>Omim: 312865</u> Human
SwissProt: 015266 Human
Unigene: 105932 Human
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