

# Rabbit Anti-FAM126A antibody

# SL11554R

<b>Product Name:</b>	FAM126A
Chinese Name:	髓鞘缺陷相关蛋白抗体
Alias:	Hyccin; Down regulated by Ctnnb1 a; Down regulated by CTNNB1 protein A; Down-regulated by CTNNB1 protein A; DRCTNNB1A; FAM126A; Family with sequence similarity 126 member A; HCC; HLD5; HYCC1; HYCCI_HUMAN antibody Hyccin; Protein FAM126A.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Dog, Pig, Horse, Rabbit,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-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:	58kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human Hyccin:1-100/521
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:	<u>PubMed</u>
Product Detail:	Hyccin is a 521 amino acid cytoplasmic protein that is widely expressed with highest levels found in heart, brain, placenta, spleen and testis. Belonging to the FAM126 family, hyccin may play a role in the ∫-catenin/Lef signaling pathway. Hyccin is likely involved in the process of myelination of the central and peripheral nervous system.

Defects in the gene encoding hyccin are the cause of leukodystrophy hypomyelinating type 5 (HLD5), which is characterized by congenital cataract, progressive neurologic impairment and diffuse myelin deficiency. Individuals affected by HLD5 experience progressive pyramidal and cerebellar dysfunction along with muscle weakness in the lower limbs. Hyccin exists as two alternatively spliced isoforms and is encoded by a gene located on human chromosome 7.

#### Function:

May have a role in the beta-catenin/Lef signaling pathway. May have a role in the process of myelination of the central and peripheral nervous system.

#### **Subcellular Location:**

Cytoplasm. Membrane. According to PubMed:10910037, it is mainly cytoplasmic while according to PubMed:16951682, it is a membrane protein.

## Tissue Specificity:

Widely expressed. Highest levels in heart, brain, placenta, spleen and testis.

#### DISEASE:

Defects in FAM126A are the cause of leukodystrophy hypomyelinating type 5 (HLD5) [MIM:610532]. This disorder is characterized by congenital cataract, progressive neurologic impairment, and diffuse myelin deficiency. Affected individuals experience progressive pyramidal and cerebellar dysfunction, muscle weakness and wasting prevailingly in the lower limbs. Mental deficiency ranges from mild to moderate.

#### Similarity:

Belongs to the FAM126 family.

# SWISS:

O9BYI3

#### Gene ID:

84668

### Database links:

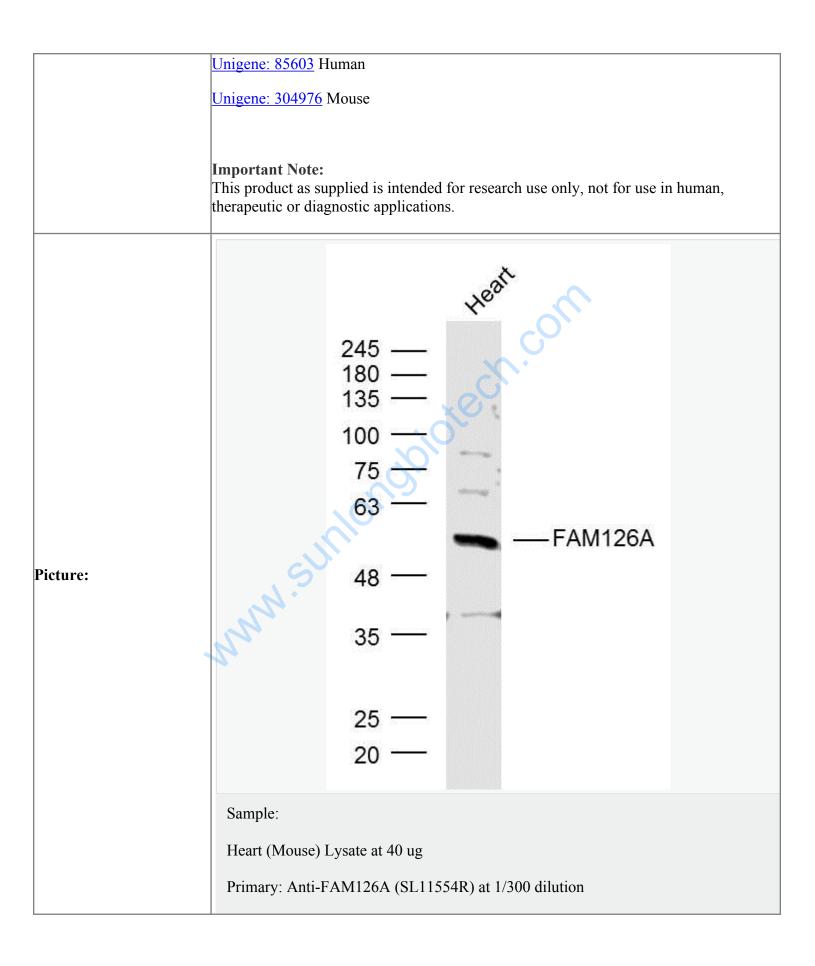
Entrez Gene: 84668 Human

Entrez Gene: 84652 Mouse

Omim: 610531 Human

SwissProt: Q9BYI3 Human

SwissProt: O6P9N1 Mouse



Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 58 kD
Observed band size: 58 kD

