abcam

Product datasheet

Recombinant Human SDHD protein ab116859

1 Image

Description

Product name Recombinant Human SDHD protein

Expression system Wheat germ
Accession O14521

Protein length Full length protein

Animal free No

Nature Recombinant

Species Human

Sequence MAVLWRLSAVCGALGGRALLLRTPVVRPAHISAFLQDRPI

PEWCGVQHIH

LSPSHHSGSKAASLHWTSERVVSVLLLGLLPAAYLNPCS

AMDYSLAAALT

LHGHWGLGQVVTDYVHGDALQKAAKAGLLALSALTFAGL

CYFNYHDVGIC KAVAMLWKL

Predicted molecular weight 44 kDa including tags

Amino acids 1 to 159

Specifications

Our Abpromise guarantee covers the use of ab116859 in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications ELISA

SDS-PAGE Western blot

Form Liquid

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

Constituents: 0.3% Glutathione, 0.79% Tris HCI

General Info

Function

Membrane-anchoring subunit of succinate dehydrogenase (SDH) that is involved in complex II of the mitochondrial electron transport chain and is responsible for transferring electrons from succinate to ubiquinone (coenzyme Q).

Pathway

Carbohydrate metabolism; tricarboxylic acid cycle.

Involvement in disease

Defects in SDHD are a cause of hereditary paragangliomas type 1 (PGL1) [MIM:168000]; also known as familial non-chromaffin paragangliomas type 1. Paragangliomas refer to rare and mostly benign tumors that arise from any component of the neuroendocrine system. PGL1 is a rare autosomal dominant disorder which is characterized by the development of mostly benign, highly vascular, slowly growing tumors in the head and neck. In the head and neck region, the carotid body is the largest of all paraganglia and is also the most common site of the tumors. Penetrance of PGL1 is incomplete when the disease is transmitted through fathers. No disease phenotype is transmitted maternally.

Defects in SDHD are a cause of susceptibility to pheochromocytoma (PCC) [MIM:171300]. A catecholamine-producing tumor of chromaffin tissue of the adrenal medulla or sympathetic paraganglia. The cardinal symptom, reflecting the increased secretion of epinephrine and norepinephrine, is hypertension, which may be persistent or intermittent.

Defects in SDHD may be a cause of susceptibility to intestinal carcinoid tumor (ICT) [MIM:114900]. A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. Defects in SDHD are a cause of paraganglioma and gastric stromal sarcoma (PGGSS) [MIM:606864]; also called Carney-Stratakis syndrome. Gastrointestinal stromal tumors may be sporadic or inherited in an autosomal dominant manner, alone or as a component of a syndrome associated with other tumors, such as in the context of neurofibromatosis type 1 (NF1). Patients have both gastrointestinal stromal tumors and paragangliomas. Susceptibility to the tumors was

Defects in SDHD are a cause of Cowden-like syndrome (CWDLS) [MIM:612359]. Cowden-like syndrome is a cancer predisposition syndrome associated with elevated risk for tumors of the breast, thyroid, kidney and uterus.

inherited in an apparently autosomal dominant manner, with incomplete penetrance.

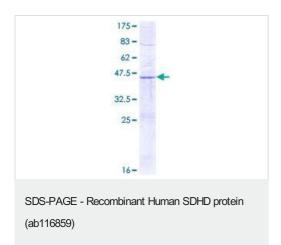
Sequence similarities

Belongs to the CybS family.

Cellular localization

Mitochondrion inner membrane.

Images



12.5% SDS-PAGE showing ab116859 at approximately 43.56kDa and stained with Coomassie Blue.

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