# abcam

## Product datasheet

# Recombinant Human SDHD protein ab116859

### 1 图像

描述

产品名称 重组人SDHD蛋白

**表达系**统 Wheat germ

Accession <u>O14521</u>

**蛋白长度** Full length protein

无动物成分 No

**性**质 Recombinant

**种属** Human

序列 MAVLWRLSAVCGALGGRALLLRTPVVRPAHISAFLQDRPIPE

WCGVQHIH

LSPSHHSGSKAASLHWTSERVVSVLLLGLLPAAYLNPCSAMD

**YSLAAALT** 

 $\verb|LHGHWGLGQVVTDYVHGDALQKAAKAGLLALSALTFAGLCYF|$ 

NYHDVGIC KAVAMLWKL

预**测分子量** 44 kDa including tags

**氨基酸** 1 to 159

#### 技术指标

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.

应**用** ELISA

SDS-PAGE

Western blot

形式 Liquid

制备和贮存

稳**定性和存储** Shipped on dry ice. Upon delivery aliquot and store at -80℃. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.3% Glutathione, 0.79% Tris HCI

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#### 常规信息

功能

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).

通路

Carbohydrate metabolism; tricarboxylic acid cycle.

疾病相关

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

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

breast, thyroid, kidney and uterus.

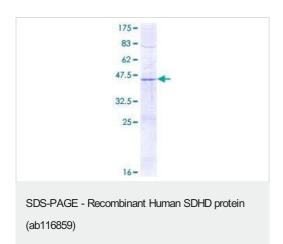
细胞定位

序列相似性

Mitochondrion inner membrane.

Belongs to the CybS family.

图片



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

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