abcam

Product datasheet

Recombinant Human SDHD protein ab116859

画像数1

製品の詳細

製品名 Recombinant Human SDHD protein

 発現系
 Wheat germ

 アクセッション番号
 O14521

タンパク質長 Full length protein

Animal free No

由来 Recombinant

生物種 Human

配列 MAVLWRLSAVCGALGGRALLLRTPVVRPAHISAFLQDRPIPE

WCGVQHIH

LSPSHHSGSKAASLHWTSERVVSVLLLGLLPAAYLNPCSAMD

YSLAAALT

 $\verb|LHGHWGLGQVVTDYVHGDALQKAAKAGLLALSALTFAGLCYF| \\$

NYHDVGIC KAVAMLWKL

予測される分子量 44 kDa including tags

領域 1 to 159

特性

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.

アプリケーション ELISA

SDS-PAGE Western blot

製品の状態 Liquid

前処理および保存

保存方法および安定性 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

関連情報

機能

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).

パスウェイ

Carbohydrate metabolism; tricarboxylic acid cycle.

関連疾患

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.

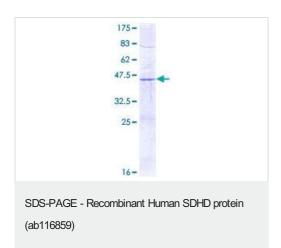
配列類似性

Belongs to the CybS family.

細胞内局在

Mitochondrion inner membrane.

画像



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

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