

# Rabbit Anti-FOXRED1 antibody

# SL13209R

Product Name:	FOXRED1
Chinese Name:	单Transmembrane proteinFOXRED1抗体
Alias:	FAD dependent oxidoreductase domain containing 1; FAD dependent oxidoreductase domain containing protein 1; FAD-dependent oxidoreductase domain-containing protein 1; FOXRED 1; FOXRED 1; FP634; FXRD1 HUMAN; H17.
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:	54kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	lmg/ml
immunogen:	KLH conjugated synthetic peptide derived from human FOXRED1:251-350/486
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:	FOXRED1 is a 486 amino acid single-pass membrane protein. Utilizing FAD as a cofactor, FOXRED1 may act as a chaperone protein essential for the function of mitochondrial complex I. Mutations to FOXRED1 may result in mitochondrial complex I deficiency (MT-C1D), which results in a wide range of clinical maladies from lethal neonatal disease to adult onset neurodegenerative disorders. Common phenotypes of

MT-C1D include cardiomyopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. FOXRED1 exists as three alternatively spliced isoforms and is encoded by a gene mapping to human chromosome 11q24.2. With approximately 135 million base pairs and 1,400 genes, chromosome 11 makes up around 4% of human genomic DNA and is considered a gene and disease association dense chromosome.

### **Subcellular Location:**

Membrane; Single-pass membrane protein (Potential).

#### DISEASE:

Defects in FOXRED1 are a cause of mitochondrial complex I deficiency (MT-C1D) [MIM:252010]. A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease.

# **SWISS:**

Q96CU9

## Gene ID:

55572

#### Database links:

Entrez Gene: 55572Human

Entrez Gene: 235169Mouse

GenBank: NP 060017.1Human

Omim: 613622Human

SwissProt: Q5EA45Cow

SwissProt: Q4R510Cynomolgus Monkey

SwissProt: Q96CU9Human

SwissProt: Q3TQB2Mouse

<u>Unigene: 317190</u>Human

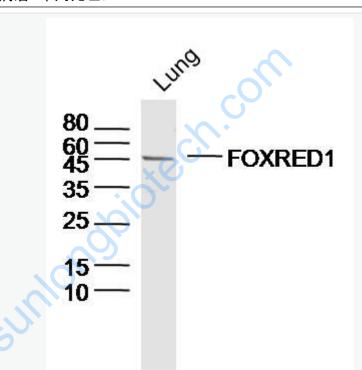
Unigene: 138512Mouse

#### **Important Note:**

This product as supplied is intended for research use only, not for use in human,

therapeutic or diagnostic applications.

Leigh综合征的发生率占新生儿的1/40,000.具有不同的基因类型,但临床具有共性特点,一般发病在1岁或以后,表现为肌张力减退,发作性呕吐,共济失调,舞蹈徐动症和过度通气,脑病表现为丧失语言发育能力,运动异常表现为痉挛性运动和异常呼吸节律,出现脑干或基底节损害体征和听力丧失,小脑损害导致共济失调,眼震和张力失常.眼科症状表现为视力丧失和眼肌麻痹.出现亚临床的周围神经病,出现神经传导速度减慢45%.临床体征可以在感染或Diabetes后出现.病程进展出现运动或智能减退.常在发病后2年内死亡.



Picture:

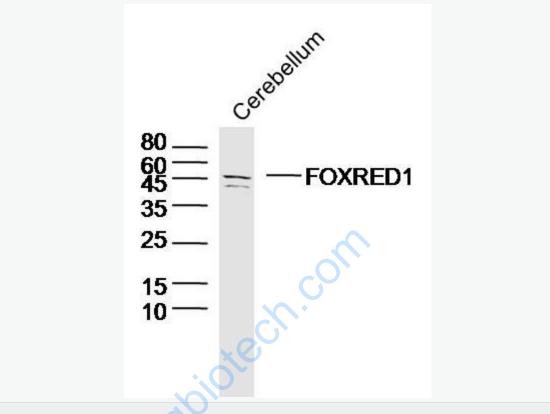
Sample: Lung (Mouse) Lysate at 40 ug

Primary: Anti-FOXRED1 (SL13209R) at 1/300 dilution

Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution

Predicted band size: 54 kD

Observed band size: 50 kD



Sample: Cerebellum (Mouse) Lysate at 40 ug

Primary: Anti-FOXRED1 (SL13209R) at 1/300 dilution

Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution

Predicted band size: 54 kD

Observed band size: 54 kD