

罗伯茨综合征相关蛋白抗体

产品货号： mlR14632

英文名称： ESCO2

中文名称： 罗伯茨综合征相关蛋白抗体

别名： CTF7, S. CEREVISIAE, HOMOLOG OF, 2; ECO1 homolog 2; ECO1, S. CEREVISIAE, HOMOLOG OF, 2; EFO2; ESO1, S. POMBE, HOMOLOG OF, 2; ESTABLISHMENT FACTOR ORTHOLOG 2; EFO2; Establishment of cohesion 1 homolog 2; Establishment of cohesion 1 homolog 2 (S. cerevisiae); N acetyltransferase ESCO2; RBS; Roberts syndrome.

研究领域： 细胞生物 信号转导 表观遗传学

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse,

产品应用： WB=1:500-2000 ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500

(石蜡切片需做抗原修复)

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分子量： 68kDa

细胞定位： 细胞核

性状： Lyophilized or Liquid

浓度： 1mg/ml

免疫原： KLH conjugated synthetic peptide derived from human ESCO2:101-200/601

亚型： IgG

纯化方法： affinity purified by Protein A

储存液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： 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： PubMed

产品介绍： This gene encodes a protein that may have acetyltransferase activity and may be required for the establishment of sister chromatid cohesion during the S phase of mitosis. Mutations in this gene have been associated with Roberts syndrome. [provided by RefSeq, Jul 2008]

Function:

ESCO2 is an acetyltransferase required for the establishment of sister chromatid cohesion, and couples the processes of cohesion and DNA replication to ensure that only sister chromatids become paired together. In contrast to the structural cohesins, the deposition and establishment factors are required only during S phase. Defects in ESCO2 are the cause of Roberts syndrome (RBS), a rare autosomal recessive disorder characterized by pre- and postnatal growth retardation, microcephaly, bilateral cleft lip and palate, and mesomelic symmetric limb reduction. Severely affected infants may be stillborn or die shortly after birth. RBS chromosomes have a lack of cohesion involving the heterochromatic C-banding regions around centromeres and the distal portion of the long arm of the Y chromosome (known as premature centromere separation, heterochromatin repulsion or puffing, or RS effect). Defects in ESCO2 are also the cause of SC phocomelia syndrome, also known as SC pseudothalidomide syndrome. SC phocomelia syndrome has a milder phenotype than RBS, with a lesser degree of symmetric limb reduction and additionally includes flexion contractures of various joints, midfacial hemangioma, hypoplastic cartilage of ears and nose, scant silvery-blond hair, and cloudy corneae. Although microcephaly is present, mental retardation may be mild and survival into adulthood is common.

Subcellular Location:

Nuclear

SWISS:

Q56N19

Gene ID:

157570

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

产品图片

