abcam

Product datasheet

Recombinant Human CSB protein ab114506

1 图像

描述

产品名称	重组人CSB蛋白
表达系统	Wheat germ
Accession	Q03468-1
蛋白长度	Protein fragment
无动物成分	No
性质	Recombinant
种属	Human
序列	RARNHLILPERLESESGHLQEASALLPTTEHDDLLVEMRNFIAFQAHTDG QASTREILQEFESKLSASQSCVFRELLRNLCTFHRTSGGEGIWKLKPEYC
预测分子量	37 kDa including tags
氨基酸	1394 to 1493

技术指标

Our **Abpromise guarantee** covers the use of **ab114506** 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

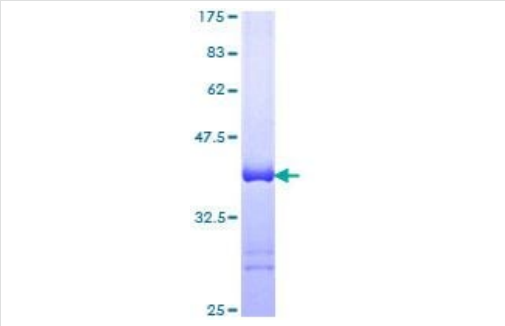
制备和贮存

稳定性和存储	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl

常规信息

功能	Essential factor involved in transcription-coupled nucleotide excision repair which allows RNA polymerase II-blocking lesions to be rapidly removed from the transcribed strand of active genes. Upon DNA-binding, it locally modifies DNA conformation by wrapping the DNA around itself, thereby modifying the interface between stalled RNA polymerase II and DNA. It is required for transcription-coupled repair complex formation. It recruits the CSA complex (DCX(ERCC8) complex), nucleotide excision repair proteins and EP300 to the at sites of RNA polymerase II-blocking lesions.
疾病相关	Defects in ERCC6 are the cause of Cockayne syndrome type B (CSB) [MIM:133540]. Cockayne syndrome is a rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. Defects in ERCC6 are the cause of cerebro-oculo-facio-skeletal syndrome type 1 (COFS1) [MIM:214150]; also known as COFS syndrome or Pena-Shokeir syndrome type 2. COFS is a degenerative autosomal recessive disorder of prenatal onset affecting the brain, eye and spinal cord. After birth, it leads to brain atrophy, hypoplasia of the corpus callosum, hypotonia, cataracts, microcornea, optic atrophy, progressive joint contractures and growth failure. Facial dysmorphism is a constant feature. Abnormalities of the skull, eyes, limbs, heart and kidney also occur. Defects in ERCC6 are a cause of De Sanctis-Cacchione syndrome (DSC) [MIM:278800]; also known as xerodermic idiocy. DSC is an autosomal recessive syndrome consisting of xeroderma pigmentosum associated with mental retardation, retarded growth, gonadal hypoplasia and sometimes neurologic complications. Note=A genetic variation in the 5-prime flanking region of ERCC6 has been shown to be associated with susceptibility to age-related macular degeneration. Defects in ERCC6 are a cause of UV-sensitive syndrome (UVS) [MIM:600630]. UVS is a rare autosomal recessive disorder characterized by photosensitivity and mild freckling but without neurological abnormalities or skin tumors.
序列相似性	Belongs to the SNF2/RAD54 helicase family. Contains 1 helicase ATP-binding domain. Contains 1 helicase C-terminal domain.
结构域	A C-terminal ubiquitin-binding domain (UBD) is essential for transcription-coupled nucleotide excision repair to proceed.
翻译后修饰	Phosphorylated upon DNA damage, probably by ATM or ATR. Ubiquitinated at the C-terminus. Ubiquitination by the CSA complex leads to ERCC6 proteasomal degradation in a UV-dependent manner.
细胞定位	Nucleus.

图片



SDS-PAGE - Recombinant Human CSB protein (ab114506)

ab114506 analysed by 12.5% SDS-PAGE and stained with Coomassie Blue.

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