abcam

Product datasheet

Human IRF6 peptide ab23204

Overview

Product name Human IRF6 peptide

Description

Nature Synthetic

Amino Acid Sequence

Species Human

Sequence C-TPSMQLPPALPPQ

Amino acids 455 to 504

Specifications

Our Abpromise guarantee covers the use of **ab23204** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications Blocking - Blocking peptide for Anti-IRF6 antibody (ab10925)

Form Liquid

Preparation and Storage

Stability and Storage Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.

General Info

Function Probable DNA-binding transcriptional activator. Key determinant of the keratinocyte proliferation-differentiation switch involved in appropriate epidermal development (By similarity). Plays a role in regulating mammary epithelial cell proliferation.

Tissue specificity Expressed in normal mammary epithelial cells. Expression is reduced or absent in breast carcinomas.

Involvement in disease Defects in IRF6 are a cause of van der Woude syndrome (VWS) [MIM:119300]; also known as lip-pit syndrome (LPS). It is an autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. Penetrance is incomplete. Van der Woude and

	popliteal pterygium syndrome are allelic disorders. Defects in IRF6 are the cause of popliteal pterygium syndrome (PPS) [MIM:119500]. PPS is an autosomal dominant developmental disorder characterized by cleft lip and/or cleft palate, and skin and genital anomalies. Penetrance is incomplete and expressivity is variable. It shows orofacial phenotypic similarities with van der Woude syndrome. Van der Woude and popliteal pterygium syndrome are allelic disorders. Genetic variation in IRF6 is associated with non-syndromic orofacial cleft type 6 (OFC6) [MIM:608864]; also called non-syndromic cleft lip with or without cleft palate 6. Non-syndromic orofacial cleft is a common birth defect consisting of cleft lips with or without cleft palate. Cleft lips are associated with cleft palate in two-third of cases. A cleft lip can occur on one or both sides and range in severity from a simple notch in the upper lip to a complete opening in the lip extending into the floor of the nostril and involving the upper gum.
Sequence similarities	Belongs to the IRF family. Contains 1 IRF tryptophan pentad repeat DNA-binding domain.
Post-translational modifications	Phosphorylated. Phosphorylation status depends on the cell cycle and is a signal for ubiquitination and proteasome-mediated degradation.
Cellular localization	Nucleus. Cytoplasm. Translocates to nucleus in response to an activating signal.

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