

Product datasheet

Human PAX3 peptide ab23270

Overview

Product name Human PAX3 peptide

Description

Nature Synthetic

Amino Acid Sequence

Species Human

Sequence TTLAGAVPRMM-C

Amino acids 2 to 12

Specifications

Our [Abpromise guarantee](#) covers the use of **ab23270** 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-PAX3 antibody ([ab15717](#))

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 transcription factor associated with development of alveolar rhabdomyosarcoma.

Involvement in disease Defects in PAX3 are the cause of Waardenburg syndrome type 1 (WS1) [MIM:193500]. WS1 is an autosomal dominant disorder characterized by wide bridge of nose owing to lateral displacement of the inner canthus of each eye (dystopia canthorum), pigmentary disturbances such as frontal white blaze of hair, heterochromia of irides, white eyelashes, leukoderma and sensorineural deafness. The syndrome shows variable clinical expression and some affected individuals do not manifest hearing impairment.

Defects in PAX3 are the cause of Waardenburg syndrome type 3 (WS3) [MIM:148820]; also

known as Klein-Waardenburg syndrome or Waardenburg syndrome with upper limb anomalies or white forelock with malformations. WS3 is a very rare autosomal dominant disorder, which shares many of the characteristics of WS1. Patients additionally present with musculoskeletal abnormalities.

Defects in PAX3 are the cause of craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]. CDHS is thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness.

Defects in PAX3 are a cause of rhabdomyosarcoma type 2 (RMS2) [MIM:268220]. It is a form of rhabdomyosarcoma, a highly malignant tumor of striated muscle derived from primitive mesenchymal cells and exhibiting differentiation along rhabdomyoblastic lines.

Rhabdomyosarcoma is one of the most frequently occurring soft tissue sarcomas and the most common in children. It occurs in four forms: alveolar, pleomorphic, embryonal and botryoidal rhabdomyosarcomas. Note=A chromosomal aberration involving PAX3 is found in rhabdomyosarcoma. Translocation (2;13)(q35;q14) with FOXO1. The resulting protein is a transcriptional activator.

Note=A chromosomal aberration involving PAX3 is a cause of rhabdomyosarcoma.

Translocation t(2;2)(q35;p23) with NCOA1 generates the NCOA1-PAX3 oncogene consisting of the N-terminus part of PAX3 and the C-terminus part of NCOA1. The fusion protein acts as a transcriptional activator. Rhabdomyosarcoma is the most common soft tissue carcinoma in childhood, representing 5-8% of all malignancies in children.

Sequence similarities

Belongs to the paired homeobox family.

Contains 1 homeobox DNA-binding domain.

Contains 1 paired domain.

Cellular localization

Nucleus.

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