

Product datasheet

Human EVC2 peptide ab22981

製品の概要

製品名 Human EVC2 peptide

製品の詳細

由来 Synthetic

アミノ酸配列

生物種 Human

配列 C-LNAKKAMRALGMD

領域 1216 to 1228

特性

Our [Abpromise guarantee](#) covers the use of **ab22981** in the following tested applications.

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

アプリケーション Blocking

製品の状態 Liquid

前処理および保存

保存方法および安定性 Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.

関連情報

関連性 EVC2 is an integral membrane protein that plays a vital role in bone formation and skeletal development. Defects in EVC2 are a cause of Ellis-van Creveld syndrome (EVC), also known as chondroectodermal dysplasia. EVC is an autosomal recessive disorder characterized by the clinical tetrad of chondrodystrophy, polydactyly, ectodermal dysplasia and cardiac anomalies. Patients manifest short-limb dwarfism, short ribs, postaxial polydactyly and dysplastic nails and teeth. Congenital heart defects, most commonly an atrioventricular septal defect, are observed in 60% of affected individuals. Defects in EVC2 are also a cause of acrofacial dysostosis Weyers type (WAD), also known as Curry-Hall syndrome. Acrofacial dysostoses are

a heterogeneous group of disorders combining limb defects with facial abnormalities. WAD is an autosomal dominant disorder characterized by dysplastic nails, postaxial polydactyly, acrofacial dysostosis, short limbs and short stature. The phenotype is milder than Ellis-van Creveld syndrome.

細胞内局在

Membrane; Multi-pass membrane protein

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