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Rabbit Anti-EVC2 antibody

SL6638R

Product Name:	EVC2
Chinese Name:	膜蛋白EVC2抗体
Alias:	Ellis van Creveld syndrome 2; LBN; Limbin; LBN_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	145kDa
Cellular localization:	The nucleuscytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human EVC2:101-200/1308<Extracellular>
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	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
Product Detail:	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.

Function:
Positive regulator of the hedgehog signaling pathway. Plays a critical role in bone formation and skeletal development.

Subunit:
Interacts with EVC.

Subcellular Location:
Cell membrane; Single-pass type I membrane protein. Cytoplasm, cytoskeleton, cilium basal body. Cell projection, cilium. Cell projection, cilium membrane. Nucleus.

Tissue Specificity:
Found in the heart, placenta, lung, liver, skeletal muscle, kidney and pancreas.

DISEASE:
Defects in EVC2 are a cause of Ellis-van Creveld syndrome (EVC) [MIM:225500]; 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 a cause of acrofacial dysostosis Weyers type (WAD) [MIM:193530]; 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.

SWISS:
Q86UK5

Gene ID:
132884

Database links:

Entrez Gene: 132884Human

Entrez Gene: 68525Mouse

Omim: 607261Human

SwissProt: Q86UK5Human

	SwissProt: Q8K1G2Mouse Unigene: 87306Human **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.