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Rabbit Anti-EVC1 antibody

SL11282R

Product Name:	EVC1
Chinese Name:	软骨外胚层发育不良相关蛋白抗体
Alias:	Ellis van Creveld syndrome DWF 1; DWF1; Ellis van Creveld syndrome protein; Ellis-van Creveld syndrome; EVC; EVC1; EVC 1; EVC-1; EVCL; MGC105107; EVC_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,
Applications:	ELISA=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:	112kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human EVC1:251-350/408
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:	EVC is an autosomal skeletal dysplasia caused by mutations in the EVC and EVC2 genes. Found in developing ribs, heart, kidney and lung, the EVC gene is responsible for normal development of the face, limbs, teeth and nails. The protein expressed by the EVC gene is an intracellular component of the hedgehog signal pathway that contains a leucine zipper and transmembrane domain. Defects in the EVC gene can lead to short-

limb dwarfism, ectodermal dysplasia and cardiac anomalies such as irregular atrioventricular septum development. Additionally, the EVC gene has been implicated in Weyers acrodental dysostosis, an autosomal dominant disease characterized by facial abnormalities and limb defects.

Function:
Ellis van Creveld syndrome 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 this protein are a cause of Ellis van Creveld syndrome (EVC).

Subunit:
Interacts with EVC2 (By similarity).

Subcellular Location:
Membrane; Single pass membrane protein

Tissue Specificity:
Found in the developing vertebral bodies, ribs, upper and lower limbs, heart, kidney, lung.

DISEASE:
Defects in EVC 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 EVC 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:
P57679

Gene ID:
2121

Database links:

	Entrez Gene: 2121Human Omim: 225500Human Omim: 604831Human SwissProt: P57679Human **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.