

Connexin 43 rabbit-FC recombinant protein

Catalog No :	YD3140
Reactivity :	Human;
Purity :	>90% as determined by SDS-PAGE
Gene Name :	Connexin 43 / GJA1
Protein Name :	Gap junction alpha-1 protein
Sequence :	Amino acid:229-382,with rabbit FC tag.
Human Gene Id :	2697
Human Swiss Prot No :	P17302
Formulation :	Phosphate-buffered solution
Source :	Mammalian cells
Storage Stability :	-15°C to -25°C/1 year(Avoid freeze / thaw cycles)
Background :	<p>This gene is a member of the connexin gene family. The encoded protein is a component of gap junctions, which are composed of arrays of intercellular channels that provide a route for the diffusion of low molecular weight materials from cell to cell. The encoded protein is the major protein of gap junctions in the heart that are thought to have a crucial role in the synchronized contraction of the heart and in embryonic development. A related intronless pseudogene has been mapped to chromosome 5. Mutations in this gene have been associated with oculodentodigital dysplasia, autosomal recessive craniometaphyseal dysplasia and heart malformations. [provided by RefSeq, May 2014],</p>
Function :	<p>caution:PubMed:11741837 reported 2 mutations (Phe-11 and Ala-24) linked to non-syndromic autosomal recessive deafness (DFNBG). These mutations have subsequently been shown (PubMed:12457340) to involve the pseudogene of connexin-43 located on chromosome 5.,caution:PubMed:7715640 reported a mutation Pro-364 linked to congenital heart diseases. This was later shown (PubMed:8873667) to be an artifact.,disease:Defects in GJA1 are a cause of hypoplastic left heart syndrome (HLHS) [MIM:241550]. HLHS refers to the abnormal development of the left-sided cardiac structures, resulting in obstruction</p>

to blood flow from the left ventricular outflow tract. In addition, the syndrome includes underdevelopment of the left ventricle, aorta, and aortic arch, as well as mitral atresia or stenosis.,disease:Defects in GJA1 are the cause of autosomal dominant oculodentodigital dysplasia (ODDD) [MIM:164200]; al

Subcellular Location :

Cell membrane ; Multi-pass membrane protein . Cell junction, gap junction . Endoplasmic reticulum . Localizes at the intercalated disk (ICD) in cardiomyocytes and the proper localization at ICD is dependent on TMEM65. .

Expression :

Expressed in the heart and fetal cochlea.

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