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# MASS1/GPR98 Rabbit pAb

MASS1/GPR98 Rabbit pAb
Catalog # AP57215

## Product Information

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| **Application** | IHC-P, IHC-F, IF |
| **Primary Accession** | Q8WXG9 |
| **Predicted** | Rat, Human, Mouse, Horse, Sheep |
| **Host** | Rabbit |
| **Clonality** | Polyclonal |
| **Calculated MW** | 693069 |
| **Physical State** | Liquid |
| **Immunogen** | KLH conjugated synthetic peptide derived from human MASS1/GPR98 |
| **Epitope Specificity** | 2451-2550/6306 |
| **Isotype** | IgG |
| **Purity** | affinity purified by Protein A |
| **Buffer** | 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. |
| **SUBCELLULAR LOCATION** | Cell membrane. |
| **DISEASE** | Defects in GPR98 are the cause of Usher syndrome type 2C (USH2C) [MIM:605472]. USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1), Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH2 is characterized by congenital mild hearing impairment with normal vestibular responses. Defects in GPR98 may be a cause of familial febrile convulsions type 4 (FEB4) [MIM:604352]; also known as familial febrile seizures 4. Febrile convulsions are seizures associated with febrile episodes in childhood without any evidence of intracranial infection or defined pathologic or traumatic cause. It is a common condition, affecting 2-5% of children aged 3 months to 5 years. The majority are simple febrile seizures (generally defined as generalized onset, single seizures with a duration of less than 30 minutes). Complex febrile seizures are characterized by focal onset, duration greater than 30 minutes, and/or more than one seizure in a 24 hour period. The likelihood of developing epilepsy following simple febrile seizures is low. Complex febrile seizures are associated with a moderately increased incidence of epilepsy. |
| **Important Note** | This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications. |
| **Background Descriptions** | MASS1 (for monogenic audiogenic seizure susceptibility 1) is one of the largest known GPCRs and is therefore referred to as Very Large G protein-coupled receptor 1 (VLGR1) (1,2). MASS1 is a large, calcium-binding GPCR expressed in the central nervous system and the eye (2,3). MASS1 has a large ectodomain containing multiple calcium exchanger beta repeats that resemble regulatory domains of sodium-calcium exchanger proteins (3). The human MASS1 gene maps to chromosome 5q14 and encodes a 1967 amino acid protein (1,2,4). The MASS1 gene has been linked to the autosomal recessive inheritance of general epilepsy in Frings mice that have seizures in response to loud noises (5). |

## Additional Information

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| **Gene ID** | 84059 |
| **Other Names** | Adhesion G-protein coupled receptor V1, ADGRV1, 3.4.-.-, ADGRV1 (HGNC:17416) |
| **Dilution** | IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500 |
| **Storage** | Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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. |

## Protein Information

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| **Name** | ADGRV1 ( HGNC:17416) |
| **Function** | G protein-coupled receptor which has an essential role in the development of hearing and vision. Couples to G-alpha(i)-proteins, GNAI1/2/3, G-alpha(q)-proteins, GNAQ, as well as G-alpha(s)-proteins, GNAS, inhibiting adenylate cyclase (AC) activity and cAMP production. Required for the hair bundle ankle formation, which connects growing stereocilia in developing cochlear hair cells of the inner ear. In response to extracellular calcium, activates kinases PKA and PKC to regulate myelination by inhibiting the ubiquitination of MAG, thus enhancing the stability of this protein in myelin-forming cells of the auditory pathway. In retina photoreceptors, the USH2 complex is required for the maintenance of periciliary membrane complex that seems to play a role in regulating intracellular protein transport. Involved in the regulation of bone metabolism. |
| **Cellular Location** | Cell membrane {ECO:0000250\|UniProtKB:Q8VHN7}; Multi-pass membrane protein {ECO:0000250\|UniProtKB:Q8VHN7}. Cell projection, stereocilium membrane {ECO:0000250\|UniProtKB:Q8VHN7} Photoreceptor inner segment {ECO:0000250\|UniProtKB:Q8VHN7} Note=Localizes at the ankle region of the stereocilia. In photoreceptors, localizes at a plasma membrane microdomain in the apical inner segment that surrounds the connecting cilia called periciliary membrane complex. {ECO:0000250\|UniProtKB:Q8VHN7} |
| **Tissue Location** | Expressed at low levels in adult tissues. |

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