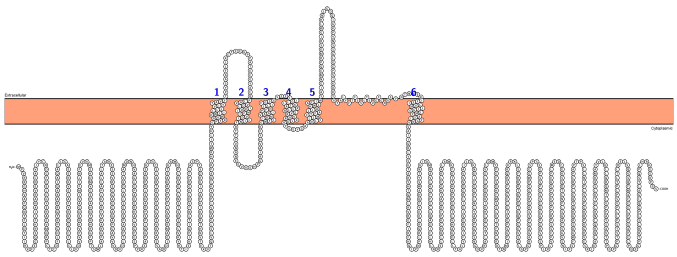
Potassium voltage-gated channel subfamily H member 2

Organism: Homo sapiens (Human) | **Gene names:** KCNH2, ERG, ERG1, HERG



Entry: Q12809

Mass: 126.655 Da

Transmembrane: 6

Subcellular location: Cell membrane {ECO:0000269|PubMed:18559421, ECO:0000269|PubMed:19412172, ECO:0000269|PubMed:26363003}, Multi-pass membrane protein {ECO:0000269|PubMed:18559421, ECO:0000269|PubMed:19412172}.

Cofactor: -

Extinction coefficient: 0.862

Isoelectric Point: 8.2

PubMed ID: 8159766, 9600240, 11374908, 12431979, 18559421, 19412172, 12853948, 9351462, 9351446, 10790218, 15489334, 9765245, 12063277, 10837251, 9230439, 10219239, 16361248, 19690332, 23186163, 25281747, 26363003, 27916661, 9845367, 7889573, 8914737, 8635257, 8877771, 9024139, 9693036, 9544837, 9452080, 10086971, 10220144, 10187793, 10517660, 10735633, 10973849, 10862094, 10753933, 11170080, 12062363, 11997281, 12442276, 12354768, 12621127, 14676148, 15051636, 15840476, 16414944, 15828882, 16922724, 19716085, 22314138

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Family: -

Function:

Pore-forming (alpha) subunit of voltage-gated inwardly rectifying potassium channel. Channel properties are modulated by cAMP and subunit assembly. Mediates the rapidly activating component of the delayed rectifying potassium current in heart (IKr) (PubMed:18559421, PubMed:26363003, PubMed:27916661). {ECO:0000269|PubMed:18559421, ECO:0000269|PubMed:26363003, ECO:0000269|PubMed:27916661}.; [Isoform A-USO]: Has no channel activity by itself, but modulates channel characteristics by forming heterotetramers with other isoforms which are retained intracellularly and undergo ubiquitin-dependent degradation. {ECO:0000269|PubMed:18559421}.; [Isoform B-USO]: Has no channel activity by itself, but modulates channel characteristics by forming heterotetramers with other isoforms which are retained intracellularly and undergo ubiquitin-dependent degradation. {ECO:0000269|PubMed:18559421}.

Data from experiment(s): Hek293 membrane pellets

DIBMA 10	No data	DIBMA 12	No data
DIBMA Glycerol	No data	DIBMA Glucosamine	No data
Amphipol 17	No data	Amphipol 18	No data
AASTY 6-45	No data	AASTY 11-45	No data
AASTY 6-50	No data	AASTY 11-50	No data
AASTY 6- 55	No data	AASTY 11- 55	No data
SMALP 502-E	No data	SMALP 140-I	No data
SMALP 300	No data	SMALP 200	No data
SMALP 140	No data	DDM	No data
DM	No data	LMNG	No data
Fos-12	No data	Digitonin-A	No data
RIPA	No data		

Data from experiment(s): Hek293 membrane pellets 1 %

DIBMA 10	No data	DIBMA 12	No data
DIBMA Glycerol	No data	DIBMA Glucosamine	No data
Amphipol 17	No data	Amphipol 18	No data
AASTY 6-45	No data	AASTY 11-45	No data
AASTY 6-50	No data	AASTY 11-50	No data
AASTY 6- 55	No data	AASTY 11- 55	No data
SMALP 502-E	No data	SMALP 140-I	No data
SMALP 300	No data	SMALP 200	No data
SMALP 140	No data	DDM	No data
DM	No data	LMNG	No data
Fos-12	No data	Digitonin-A	No data
RIPA	No data		

Involvement in disease:

Long QT syndrome 2 (LQT2) [MIM:613688]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Deafness is often associated with long QT syndrome type 2. {ECO:0000269|PubMed:10086971, ECO:0000269|PubMed:10187793, ECO:0000269|PubMed:10220144, ECO:0000269|PubMed:10517660, ECO:0000269|PubMed:10735633, ECO:0000269|PubMed:10753933, ECO:0000269|PubMed:10862094, ECO:0000269|PubMed:10973849, ECO:0000269|PubMed:11170080, ECO:0000269|PubMed:12062363, ECO:0000269|PubMed:12354768, ECO:0000269|PubMed:12442276, ECO:0000269|PubMed:12621127, ECO:0000269|PubMed:15051636, ECO:0000269|PubMed:15840476, ECO:0000269|PubMed:16361248, ECO:0000269|PubMed:16414944, ECO:0000269|PubMed:16922724, ECO:0000269|PubMed:19716085, ECO:0000269|PubMed:22314138, ECO:0000269|PubMed:27916661, ECO:0000269|PubMed:7889573, ECO:0000269|PubMed:8635257, ECO:0000269|PubMed:8877771, ECO:0000269|PubMed:8914737, ECO:0000269|PubMed:9024139, ECO:0000269|PubMed:9452080, ECO:0000269|PubMed:9544837, ECO:0000269|PubMed:9600240, ECO:0000269|PubMed:9693036}. Note=The disease is caused by variants affecting the gene represented in this entry.; Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. {ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882}. Note=The disease is caused by variants affecting the gene represented in this entry.

Binding site:

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Tissue specificity:

Highly expressed in heart and brain. Isoforms USO are frequently overexpressed in cancer cells. {ECO:0000269|PubMed:18559421}.

3D (X-ray crystallography):

X-ray crystallography (4); NMR spectroscopy (6); Electron microscopy (3)

Pharmaceutical use:

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AS sequence:

MPVRRGHVAPQNTFLDTIIRKFEGQSRKFIIANARVENCAVIYCNDGFCELCGYSRAEVMQRPCTCDFLHGPRTQRRAAAQIAQA
LLGAEERKVEIAFYRKDGSCFLCLVDVVPVKNEDGAVIMFILNFEVVMEKDMVGSPAHDTNHRGPPTSWLAPGRAKTFRLKLPA
LLALTARESSVRSGGAGGAGAPGAVVVDVDLTPAAPSSESLALDEVTAMDNHVAGLGPAEERRALVGPGSPPRSAPGQLPSPR
AHSLNPDASGSSCSLARTRSRESCASVRRASSADDIEAMRAGVLPPPPRHASTGAMHPLRSGLLNSTSDSDLVRYRTISKIPQIT
LNFVDLKGDPFLASPTSDREIIAPKIKERTHNVTEKVTQVLSLGADVLPEYKLQAPRIHRWTILHYSPFKAVWDWLILLLVIYTAVF
TPYSAAFLLKETEEGPPATECGYACQPLAVVDLIVDIMFIVDILINFRTTYVNANEEVVSHPGRIAVHYFKGWFLIDMVAAIPFDLLI
FGSGSEELIGLLKTARLLRLVRVARKLDRYSEYGAAVLFLLMCTFALIAHWLACIWYAIGNMEQPHMDSRIGWLHNLGDQIGKPY
NSSGLGGPSIKDKYVTALYFTFSSLTSVGFGNVSPNTNSEKIFSICVMLIGSLMYASIFGNVSAIIQRLYSGTARYHTQMLRVREFIR
FHQIPNPLRQRLEEYFQHAWSYTNGIDMNAVLKGFPECLQADICLHLNRSLLQHCKPFRGATKGCLRALAMKFKTTHAPPGDTL
VHAGDLLTALYFISRGSIEILRGDVVVAILGKNDIFGEPLNLYARPGKSNGDVRALTYCDLHKIHRDDLLEVLDMYPEFSDHFWSS
LEITFNLRDTNMIPGSPGSTELEGGFSRQRKRKLSFRRRTDKDTEQPGEVSALGPGRAGAGPSSRGRPGGPWGESPSSGPSSPE
SSEDEGPGRSSSPLRLVPFSSPRPPGEPPGGEPLMEDCEKSSDTCNPLSGAFSGVSNIFSFWGDSRGRQYQELPRCPAPTPSLL
NIPLSSPGRRPRGDVESRLDALQRQLNRLETRLSADMATVLQLLQRQMTLVPPAYSAVTTPGPGPTSTSPLLPVSPLPTLTLDSL
SQVSQFMACEELPPGAPELPQEGPTRRLSLPGQLGALTSQPLHRHGSDPGS

Creditnotes:

The protein visualizations are generated with the help of Protter:
Omasits, U., Ahrens, C.H., MÃ¼ller, S., Wollscheid, B. "Protter: interactive protein feature visualization and integration with experimental proteomic data". *Bioinformatics*. 2014 Mar 15; **30**(6):884-6. doi: 10.1093/bioinformatics/btt607.

IP and extinction coefficients are gathered from Protparam by ExPASy:
Gasteiger, E., Hoogland, C., Gattiker, A., Duvaud, S., Wilkins, M.R., Appel, R.D., Bairoch, A. "Protein Identification and Analysis Tools on the ExPASy Server". (In) *John M. Walker (ed): The Proteomics Protocols Handbook*, Humana Press (2005). pp. 571-607

The basic knowledge is found on UniProt:
The UniProt Consortium. "UniProt: the universal protein knowledgebase in 2021". *Nucleic Acids Res*. **49**:D1 (2021)