# ACVR2 (Activin Receptor Type 2)

## Identity

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|---|---|
| Other names | **ACVRII**<br>**ACVR2A**<br>**Activin Receptor**<br>**Type 2A**<br>**Activin Receptor A Type 2**<br>**T ACTRII**<br>**ActR-II** |
| Hugo | **ACVR2A** |
| Location | 2q22.3 |
| Local_order | Genes flanking ACVR2A, in centromere to telomere direction on 2q22, are:<br>PAPBPCP2 2q22.3 polyadenylate binding protein, cytoplasmic, pseudogene 2.<br>ACVR2 2q22.3 activin receptor type IIA.<br>ORC4L 2q23.1 origin recognition complex, subunit 4. |

## DNA/RNA

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| Description | ACVR2 gene spans a region of 85,796 bp and has 11 exons. Exon lengths are 180, 208, 110, 155, 144, 144, 146, 115, 139, 131 and 3745 base pairs. Exon 10 contains a polyadenine tract that may be mutated in microsatellite unstable cells. |
| Transcription | The transcript is 5217 base pairs. |

## Protein

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| Description | ACVR2 is a member of the transforming growth factor beta (TGF-b) receptor family. It is a 70-75kDA protein consisting of 513 amino acids. It is a transmembrane receptor for activin, with a cysteine-rich extracellular ligand-binding domain, a single pass transmembrane domain, and an intracellular domain with constitutive serine/threonine kinase activity.<br>Upon binding activin, ACVR2 associates with and phosphorylates ACVR1.<br>ACVR1, in turn, phosphorylates Smad2 and/or Smad3. Phosphorylated Smad2 and Smad3 associate with Smad4, translocate to the nucleus, and regulate gene expression.<br>There may be other non-Smad pathways in activin signal transduction. These include the RhoA-ROCK-MEKK1-JNK and MEKK1-p38 pathways.<br>In addition to activin, other ligands such as myostatin, nodal, and bone morphogenetic protein 7 (BMP-7) may also bind to ACVR2 and affect signal transduction. |
| Expression | Abundant expression in multiple tissues, including skeletal muscle, stomach, heart, endometrium, testes, prostate, ovary, and neural tissues. The cell surface level of ACVR2 and ACVR2B is regulated by proteins called ARIPs (activin receptor-interacting proteins). |
| Localisation | Cell surface, spanning cytoplasmic membrane. |
| Function | Activin signaling via its receptors has roles in cell proliferation, differentiation, apoptosis, metabolism, immune response, wound repair, and endocrine function. |

## Mutations

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| Somatic | Exon 10 polyadenine tract (A8-A7). |

## Implicated in

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| **Entity** | Colon cancer |
| **Note** | Colon cancer with A8-A7 deletion in exon10. |
| Disease | Microsatellite unstable colon cancer. |

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| Prognosis | Increased tumor size. |
| Abnormal Protein | No fusion protein; truncated non-functional protein. |
| Oncogenesis | Occurs late in adenoma to carcinoma transition. |

## External links

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|---|---|
| | **Nomenclature** |
| Hugo | ACVR2A |
| GDB | ACVR2A |
| Entrez_Gene | ACVR2A 92 activin A receptor, type IIA |
| | **Cards** |
| **Atlas** | **ACVR2ID567ch2q22** |
| GeneCards | ACVR2A |
| Ensembl | ACVR2A [Search_View] ENSG00000121989 [Gene_View] |
| Genatlas | ACVR2A |
| GeneLynx | ACVR2A |
| eGenome | ACVR2A |
| euGene | 92 |
| | **Genomic and cartography** |
| GoldenPath | ACVR2A - 2q22.3 chr2:148319040-148404862 + 2q22.3 (hg18-Mar_2006) |
| Ensembl | ACVR2A - 2q22.3 [CytoView] |
| NCBI | Mapview |
| OMIM | Disease map [OMIM] |
| HomoloGene | ACVR2A |
| | **Gene and transcription** |
| Genbank | BC067416 [ ENTREZ ] |
| Genbank | BC067417 [ ENTREZ ] |
| Genbank | BC067418 [ ENTREZ ] |
| Genbank | BC069707 [ ENTREZ ] |
| Genbank | BX645435 [ ENTREZ ] |
| RefSeq | NM_001616 [ SRS ] NM_001616 [ ENTREZ ] |
| RefSeq | AC_000045 [ SRS ] AC_000045 [ ENTREZ ] |
| RefSeq | NC_000002 [ SRS ] NC_000002 [ ENTREZ ] |
| RefSeq | NT_022135 [ SRS ] NT_022135 [ ENTREZ ] |
| RefSeq | NW_921585 [ SRS ] NW_921585 [ ENTREZ ] |
| AceView | ACVR2A AceView - NCBI |
| Unigene | Hs.470174 [ SRS ] Hs.470174 [ NCBI ] HS470174 [ spliceNest ] |
| Fast-db | 11585 (alternative variants) |
| | **Protein : pattern, domain, 3D structure** |
| HPRD | 03193 |
| | **Protein Interaction databases** |
| | **Polymorphism : SNP, mutations, diseases** |
| OMIM | 102581 [ map ] |
| GENECLINICS | 102581 |
| SNP | ACVR2A [dbSNP-NCBI] |
| SNP | NM_001616 [SNP-NCI] |

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|---|---|
| SNP | ACVR2A [GeneSNPs - Utah] ACVR2A] [HGBASE - SRS] |
| HAPMAP | ACVR2A [HAPMAP] |
| COSMIC | ACVR2A [Somatic mutation (COSMIC-CGP-Sanger)] |
| HGMD | ACVR2A |
| | **General knowledge** |
| Family Browser | ACVR2A [UCSC Family Browser] |
| SOURCE | NM_001616 |
| SMD | Hs.470174 |
| SAGE | Hs.470174 |
| GO | nucleotide binding [Amigo] nucleotide binding |
| GO | magnesium ion binding [Amigo] magnesium ion binding |
| GO | receptor activity [Amigo] receptor activity |
| GO | ATP binding [Amigo] ATP binding |
| GO | cytoplasm [Amigo] cytoplasm |
| GO | integral to plasma membrane [Amigo] integral to plasma membrane |
| GO | protein amino acid phosphorylation [Amigo] protein amino acid phosphorylation |
| GO | transmembrane receptor protein serine/threonine kinase signaling pathway [Amigo] transmembrane receptor protein serine/threonine kinase signaling pathway |
| GO | spermatogenesis [Amigo] spermatogenesis |
| GO | determination of left/right symmetry [Amigo] determination of left/right symmetry |
| GO | mesoderm development [Amigo] mesoderm development |
| GO | anterior/posterior pattern formation [Amigo] anterior/posterior pattern formation |
| GO | membrane [Amigo] membrane |
| GO | transferase activity [Amigo] transferase activity |
| GO | activin receptor activity [Amigo] activin receptor activity |
| GO | growth factor binding [Amigo] growth factor binding |
| GO | manganese ion binding [Amigo] manganese ion binding |
| GO | positive regulation of activin receptor signaling pathway [Amigo] positive regulation of activin receptor signaling pathway |
| GO | sperm ejaculation [Amigo] sperm ejaculation |
| GO | penile erection [Amigo] penile erection |
| GO | protein self-association [Amigo] protein self-association |
| GO | positive regulation of erythrocyte differentiation [Amigo] positive regulation of erythrocyte differentiation |
| GO | inhibin beta-A binding [Amigo] inhibin beta-A binding |

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| GO | embryonic skeletal development [Amigo] embryonic skeletal development |
| GO | regulation of nitric-oxide synthase activity [Amigo] regulation of nitric-oxide synthase activity |
| GO | Sertoli cell proliferation [Amigo] Sertoli cell proliferation |
| KEGG | Cytokine-cytokine receptor interaction |
| KEGG | TGF-beta signaling pathway |
| PubGene | ACVR2A |
| | **Other databases** |
| | **Probes** |
| Probe | ACVR2A Related clones (RZPD - Berlin) |
| | **PubMed** |
| PubMed | **44 Pubmed reference(s) in LocusLink** |

**Bibliography**


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Medline 8395525

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Medline 14988818

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Medline 16636301

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Medline 16380996

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Medline 16648306

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Gastroenterology 2007; 132: 633-644.
Medline 17258738

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## Citation

*This paper should be referenced as such :*

**Boles S, Chau E, Jung B** . ACVR2 (Activin Receptor Type 2). Atlas Genet Cytogenet Oncol Haematol. June 2007 .
URL : http://AtlasGeneticsOncology.org/Genes/ACVR2ID567ch2q22.html