PRKAR1A gene
protein kinase cAMP-dependent type I regulatory subunit alpha

Normal Function

The PRKAR1A gene provides instructions for making one part (subunit) of an enzyme called protein kinase A. This enzyme promotes cell growth and division (proliferation). Protein kinase A is made up of four protein subunits, two of which are called regulatory subunits because they control whether this enzyme is turned on or off. The PRKAR1A gene provides instructions for making one of these regulatory subunits, called type 1 alpha. Protein kinase A remains turned off when the regulatory subunits are attached to the other two subunits of the enzyme. In order to turn on protein kinase A, the regulatory subunits must break away from the enzyme.

Health Conditions Related to Genetic Changes

Carney complex

More than 117 mutations in the PRKAR1A gene have been found to cause Carney complex. Most of these mutations result in an abnormal type 1 alpha regulatory subunit that is quickly broken down (degraded) by the cell. The lack of this regulatory subunit causes protein kinase A to be turned on more often than normal, which leads to uncontrolled cell proliferation. The signs and symptoms of Carney complex are related to the unregulated growth of cells in many parts of the body.

Chromosomal Location

Cytogenetic Location: 17q24.2, which is the long (q) arm of chromosome 17 at position 24.2

Molecular Location: base pairs 68,413,623 to 68,551,316 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

- cAMP-dependent protein kinase regulatory subunit RIalpha
- cAMP-dependent protein kinase type I-alpha regulatory chain
- CAR
- CNC1
- DKFZp779L0468
- KAP0_HUMAN
- MGC17251
- PKR1
- PPNAD1
- PRKAR1
- protein kinase A type 1a regulatory subunit
- protein kinase, cAMP-dependent, regulatory subunit type I alpha
- protein kinase, cAMP-dependent, regulatory, type I, alpha
- protein kinase, cAMP-dependent, regulatory, type I, alpha (tissue specific extinguisher 1)
- tissue-specific extinguisher 1
- TSE1

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Cyclic AMP Activates Protein Kinase A by Altering the Quaternary Structure
  https://www.ncbi.nlm.nih.gov/books/NBK22399/#A1373
- The Cell, A Molecular Approach (second edition, 2000): Regulation of Protein Kinase A
  https://www.ncbi.nlm.nih.gov/books/NBK9870/?rendertype=figure&id=A2237

Clinical Information from GeneReviews

- Carney Complex
  https://www.ncbi.nlm.nih.gov/books/NBK1286
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PRKAR1A%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PROTEIN KINASE, cAMP-DEPENDENT, REGULATORY, TYPE I, ALPHA
  http://omim.org/entry/188830

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/PRKAR1AID387.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PRKAR1A%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5573
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P10644

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19293268
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2690418/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18223213
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10973256
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http://omim.org/entry/188830

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19915019
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2805491/

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301463

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https://ghr.nlm.nih.gov/gene/PRKAR1A

Reviewed: January 2010
Published: August 17, 2020

Lister Hill National Center for Biomedical Communications
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