### EYA1 gene

**EYA transcriptional coactivator and phosphatase 1**

#### Normal Function

The *EYA1* gene provides instructions for making a protein that plays a role in regulating the activity of other genes. Based on this role, the EYA1 protein is called a transcription factor or transcription coactivator.

The EYA1 protein interacts with several other proteins, including a group known as SIX proteins, to turn on (activate) and turn off (inactivate) genes that are important for normal development. Before birth, these protein interactions appear to be essential for the normal formation of many tissues. These include the second branchial arch, which gives rise to tissues in the front and side of the neck, and the eyes, ears, and kidneys. After birth, these interactions are important for normal organ function.

#### Health Conditions Related to Genetic Changes

**Branchiootorenal/branchiootic syndrome**

At least 160 mutations in the *EYA1* gene have been identified in people with branchiootorenal (BOR) syndrome, a condition that disrupts the development of tissues in the neck and causes malformations of the ears and kidneys. *EYA1* gene mutations have also been found to cause branchiootic (BO) syndrome, which includes many of the same features as BOR syndrome except for kidney (renal) malformations. The two conditions are otherwise so similar that researchers often consider them together (BOR/BO syndrome or branchiootorenal spectrum disorders).

Many of the mutations that cause BOR/BO syndrome change the 3-dimensional structure of the EYA1 protein, which prevents it from interacting effectively with other proteins. Because these protein interactions are necessary for the activation of certain genes during embryonic development, the altered EYA1 protein impairs the normal development of many tissues before birth. The major signs and symptoms of BOR/BO syndrome result from abnormal development of the second branchial arch, the ears, and (in BOR syndrome) the kidneys.

In some cases, the same *EYA1* gene mutation causes BOR syndrome in some members of a family and BO syndrome in others. This variability might result from changes in other, unidentified genes that affect how the EYA1 protein functions in the kidneys.

**Congenital anomalies of kidney and urinary tract**
Other disorders

Several mutations in the EYA1 gene have been associated with eye abnormalities including clouding of the lens (cataracts) and clouding of the clear front surface of the eye (the cornea). These abnormalities occur without the characteristic features of BOR/BO syndrome. Researchers believe that the EYA1 gene mutations responsible for eye abnormalities have less severe effects on protein function than the mutations that underlie BOR/BO syndrome.

Chromosomal Location

Cytogenetic Location: 8q13.3, which is the long (q) arm of chromosome 8 at position 13.3

Molecular Location: base pairs 71,197,433 to 71,548,104 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BOP
- BOR
- EYA1_HUMAN
- eyes absent 1
- eyes absent homolog 1 (Drosophila)
- eyes absent, Drosophila, homolog of, 1

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Transcription Factors
  https://www.ncbi.nlm.nih.gov/books/NBK10023/#A763

Clinical Information from GeneReviews

- Branchiootorenal Spectrum Disorder
  https://www.ncbi.nlm.nih.gov/books/NBK1380
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28EYA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- EYA TRANSCRIPTIONAL COACTIVATOR AND PHOSPHATASE 1
  http://omim.org/entry/601653

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EYA1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EYA1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2138
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q99502

Sources for This Summary

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