EYA1 Gene

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EYA transcriptional coactivator and phosphatase 1

genes

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.

2. Health Conditions Related to Genetic Changes

2.1 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.

2.2 Congenital Anomalies of Kidney and Urinary Tract

2.3 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.

3. Other Names for This Gene

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

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