RBM8A Gene

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RNA binding motif protein 8A

genes

1. Normal Function

The *RBM8A* gene provides instructions for making a protein called RNA-binding motif protein 8A. This protein is believed to be involved in several important cellular functions involving protein production. These functions include helping to transport molecules called messenger RNA (mRNA), which serve as the genetic blueprint for making proteins. RNA-binding motif protein 8A likely carries mRNA molecules from the nucleus to areas of the cell where proteins are assembled. It may also be involved in controlling how the instructions in mRNA molecules are used to build proteins and in destroying mRNA that is defective or not needed.

2. Health Conditions Related to Genetic Changes

2.1. Thrombocytopenia-absent radius syndrome

Mutations in the *RBM8A* gene cause thrombocytopenia-absent radius (TAR) syndrome. This disorder is characterized by the absence of a bone called the radius in each forearm and a shortage (deficiency) of blood cells involved in clotting (platelets).

Most people with TAR syndrome have a mutation in one copy of the *RBM8A* gene and a deletion of genetic material from chromosome 1 that includes the other copy of the *RBM8A* gene in each cell. A small number of affected individuals have mutations in both copies of the *RBM8A* gene in each cell and do not have a deletion on chromosome 1. *RBM8A* gene mutations that cause TAR syndrome reduce the amount of RNA-binding motif protein 8A in cells. The deletions involved in TAR syndrome eliminate at least 200,000 DNA building blocks (200 kilobases, or 200 kb) from the long (q) arm of chromosome 1 in a region called 1q21.1. The deletion eliminates one copy of the *RBM8A* gene in each cell and the RNA-binding motif protein 8A that would have been produced from it.

People with either an *RBM8A* gene mutation and a chromosome 1 deletion or with two gene mutations have a decreased amount of RNA-binding motif protein 8A. This reduction is thought to cause problems in the development of certain tissues, but it is unknown how it causes the specific signs and symptoms of TAR syndrome.

No cases have been reported in which a deletion that includes the *RBM8A* gene occurs on both copies of chromosome 1; studies indicate that the complete loss of RNA-binding motif protein 8A is not compatible with life.

3. Other Names for This Gene

- binder of OVCA1-1
- BOV-1
- BOV-1A
- BOV-1B
- BOV-1C
- MDS014
- RBM8
- RBM8A_HUMAN
- RBM8B
- ribonucleoprotein RBM8
- ribonucleoprotein RBM8A
- RNA binding motif protein 8B
- RNA-binding motif protein 8A
- RNA-binding protein 8A
- RNA-binding protein Y14
- TAR
- Y14
- ZNRP

- ZRNP1

References

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