

## RBM8A gene

RNA binding motif protein 8A

### 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.

### Health Conditions Related to Genetic Changes

#### 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.

### 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

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of RBM8A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9939\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9939[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RBM8A%5BTIAB%5D%29+OR+%28RNA+binding+motif+protein+8A%5BTIAB%5D%29%29+OR+%28%28BOV-1%5BTIAB%5D%29+OR+%28RBM8%5BTIAB%5D%29+OR+%28RBM8B%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29%29>)

#### Catalog of Genes and Diseases from OMIM

- RNA-BINDING MOTIF PROTEIN 8A; RBM8A (<https://omim.org/entry/605313>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9939>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RBM8A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RBM8A[gene]))

### **References**

- Albers CA, Paul DS, Schulze H, Freson K, Stephens JC, Smethurst PA, Jolley JD, Cvejic A, Kostadima M, Bertone P, Breuning MH, Debili N, Deloukas P, Favier R, Fiedler J, Hobbs CM, Huang N, Hurles ME, Kiddle G, Krapels I, Nurden P, Ruivenkamp CA, Sambrook JG, Smith K, Stemple DL, Strauss G, Thys C, van Geet C, Newbury-Ecob R, Ouwehand WH, Ghevaert C. Compound inheritance of a low-frequency regulatory SNP and a rare null mutation in exon-junction complex subunit RBM8A causes TAR syndrome. *Nat Genet.* 2012 Feb 26;44(4):435-9, S1-2. doi:10.1038/ng.1083. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22366785>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3428915/>)
- Faurholm B, Millar RP, Katz AA. The genes encoding the type II gonadotropin-releasing hormone receptor and the ribonucleoprotein RBM8A in humans overlap in two genomic loci. *Genomics.* 2001 Nov;78(1-2):15-8. doi:10.1006/geno.2001.6650. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11707068>)
- Salicioni AM, Xi M, Vanderveer LA, Balsara B, Testa JR, Dunbrack RL Jr, Godwin AK. Identification and structural analysis of human RBM8A and RBM8B: two highly conserved RNA-binding motif proteins that interact with OVCA1, a candidate tumor suppressor. *Genomics.* 2000 Oct 1;69(1):54-62. doi: 10.1006/geno.2000.6315. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11013075>)

### **Genomic Location**

The *RBM8A* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

**Last updated October 1, 2012**