

# TIMM8A Gene

Subjects: Genetics & Heredity

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Translocase of inner mitochondrial membrane 8A: The TIMM8A gene provides instructions for making a protein that is found inside mitochondria, which are structures within cells that convert the energy from food into a form that cells can use.

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## 1. Normal Function

The *TIMM8A* gene provides instructions for making a protein that is found inside mitochondria, which are structures within cells that convert the energy from food into a form that cells can use. Mitochondria have two membranes, an outer membrane and an inner membrane, which are separated by a fluid-filled area called the intermembrane space. The TIMM8A protein is found in the intermembrane space, where it forms a complex (a group of proteins that work together) with a very similar protein called TIMM13. This complex transports other proteins across the intermembrane space to the mitochondrial inner membrane.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Deafness-dystonia-optic neuropathy syndrome

At least 20 mutations in the *TIMM8A* gene have been found to cause deafness-dystonia-optic neuropathy (DDON) syndrome. Most of these mutations result in the absence of functional TIMM8A protein inside the mitochondria, which prevents the formation of the TIMM8A/TIMM13 complex. Researchers believe that the lack of this complex leads to abnormal transport of proteins across the intermembrane space, although it is unclear how abnormal protein transport affects the function of the mitochondria and causes the signs and symptoms of DDON syndrome.

Some people with DDON syndrome have large DNA deletions that remove the entire *TIMM8A* gene and one end of a neighboring gene known as *BTK*. Mutations in the *BTK* gene cause X-linked agammaglobulinemia (XLA), which is characterized by an increased susceptibility to infections. Individuals with large DNA deletions that include the *TIMM8A* gene and the *BTK* gene have the signs and symptoms of both DDON syndrome and XLA.

## 3. Other Names for This Gene

- DDP
  - DDP1
  - deafness/dystonia peptide
  - DFN1
  - MGC12262
  - TIM8A\_HUMAN
  - translocase of inner mitochondrial membrane 8 homolog A
  - translocase of inner mitochondrial membrane 8 homolog A (yeast)
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