**# Gene symbol**

CHRNE

**# Full name of gene**

Acetylcholine receptor subunit epsilon

**# What does the <gene\_symbol> gene do?**

The CHRNE gene encodes a protein found in neuromuscular junctions that opens ion-conducting channels across cell membranes. It is involved in [muscle contractions, response to nicotine, synaptic transmission, and transport](http://www.uniprot.org/uniprot/Q04844#function). Variants in CHRNE are associated with the autoimmune disorder [myasthenia gravis](https://www.omim.org/entry/254200) as well as congenital myasthenic syndrome [4a](https://www.omim.org/entry/605809), [4b](https://www.omim.org/entry/616324), and [4c](https://www.omim.org/entry/608931), which causes progressive muscle weakness. Other variants reduce natural killer cell function in the immune system and are associated with [CFS](https://www.ncbi.nlm.nih.gov/pubmed/27099524).

This gene is located on chromosome 17. The protein it creates acts in your immune system and muscles.

<body part brain, immune, circularity, muscles>

<gene\_expression\_location location="D001921; D007107; D002319; D009132;>

**# What are some common mutations of <gene\_symbol>?**

There are 2 well known variants in this gene: <variant list?>

< A1074G variant view with A to G transformation>

This variant is a change at a specific point in the <gene\_symbol> gene from adenine (A) to guanine (G), resulting in reduced protein function. This substitution of a single nucleotide is known as a missense variant.

< T10927C variant view with T to C transformation>

This mutation is a change at a specific point in the <gene\_symbol> gene from thymine (T) to cytosine (C), resulting in reduced natural killer cell function. This substitution of a single nucleotide is known as a mis-sense mutation.

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<user variant viewer>

<user what does this mean>

<user gene list>

<UserGenotypeBox gene="comt">

<Genotype name=" A1074G (A;G)">

# What does this mean?

People with this variant have one copy of the A1074G variant. This substitution of a single nucleotide is known as a mis-sense mutation.

# What is the effect of this variant?

Your variant is not associated with any loss of function.

# How common is this gene in the general population?

<pie\_chart level=" A1074G (A;G)", frequency>

<frequency> of the population has a moderate loss of function

</Genotype>

<Genotype name=" A1074G (G;G)">

<line\_graph level="severe">

# What does this mean?

People with this variant have two copies of the A1074G variant. This substitution of a single nucleotide is known as a mis-sense mutation.

# What is the effect of this variant?

You are in the Severe Risk category. See below for more information

# How common is this gene in the general population?

<pie\_chart level=" A1074G (G;G)", frequency>

<frequency> of the population has a severe loss of function

<pie\_chart level=" A1074G (G;G)", frequency>

You are in the Severe Loss of Function category. See below for more information

</Genotype>

<Genotype name="A1074G (A;A)”>

# How common is this gene in the general population?

<pie\_chart level="A1074G (A;A), frequency>

Your <gene\_name> is found to have no variants. A normal gene is referred to as a "wildtype" gene.

</Genotype>

<Genotype name=" T10927C (T;C)">

<line\_graph level="moderate">

# What does this mean?

People with this variant have one copy of the T10927C variant. This substitution of a single nucleotide is known as a mis-sense mutation.

# What is the effect of this variant?

This variant is not associated with increased risk.

# How common is this gene in the general population?

<pie\_chart level="T10927C (T;C)", frequency>

<frequency> of the population has a moderate loss of function

</Genotype>

<Genotype name="T10927C (C;C)">

<line\_graph level="severe">

# What does this mean?

People with this variant have two copies of the T10927C variant. This substitution of a single nucleotide is known as a mis-sense mutation.

# What is the effect of this variant?

You are in the Severe Loss of Function category. See below for more information

# How common is this gene in the general population?

<pie\_chart level="T10927C (C;C)", frequency>

<frequency> of the population has a severe loss of function

<pie\_chart level="T10927C (C;C)", frequency>

You are in the Severe Loss of Function category. See below for more information

</Genotype>

<Genotype name="T10927C (T;T)”>

# How common is this gene in the general population?

<pie\_chart level="T10927C (T;T), frequency>

Your <gene\_name> is found to have no variants. A normal gene is referred to as a "wildtype" gene.

</Genotype>

# How sure are you?

<user gene chip graph level="user gene surity">

We have <user gene surity> confidence in the read of the gene due to the coverage of our chipset process.

</UserGenotypeBox>

# How sure are you?

<if chipset to low graph>

<level graph>

We have low confidence of this gene due to the data our chipset provides.

<else if chipset to medium graph>

<level graph>

We have medium confidence of this gene due to the data our chipset provides.

<else check chipset to high graph>

<level graph>

We have high confidence of this gene due to the data our chipset provides.

```

**# What are the effects of variants in <gene\_symbol>?**

For the vast majority of people, the overall risk associated with the common <gene\_symbol> variants is small, and do not impact treatment. It is possible that variants in this gene interact with other gene variants, which is the reason for our inclusion of this gene in the gene panel.

**<call variants with the multiple categories>**

A1074G (A;A)

**<function meter level="wildtype">**

**<efficiency level = "100%" >**

**<variant and population data>**

**# Normal Function**

The CHRNE protein enables fully functional natural killer cells (NKC) with no increased risk of CFS.

**# What should I do about this?**

No medical therapies are indicated at the moment.

A1074G (A;G)

**<function meter level="wildtype">**

**<variant and population data>**

**# Heterozygous**

The CHRNE protein enables fully functional natural killer cells (NKC) with no increased risk of CFS.

**# What should I do about this?**

No medical therapies are indicated at the moment.

A1074G (G;G)

**<function meter level="severe">**

**<variant and population data>**

**# Severe Risk**

Natural killer cells (NKC) are a type of white blood cells found in the blood, bone marrow, spleen, and lymph nodes. They kill viral infected cells and tumorous cells. CFS patients have half the cellular efficiency of the normal population with a [17% cellular death rate](https://www.ncbi.nlm.nih.gov/pubmed/27099524). The A1074G variant decreases gene expression in both the DNA and RNA, causing significant reduction in NKC activity. This variant was twice as common in [CFS patients at 62.1%.](https://www.ncbi.nlm.nih.gov/pubmed/27099524)

**# What should I do about this?**

Many dietary supplements have been found to increase or decrease natural killer cell function.

* [Resveratrol](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4855330/) stimulates the immune system in increase NKC activity, but sufficient body concentration can only be achieved through supplementation.
* [Myricetin](https://www.ncbi.nlm.nih.gov/pubmed/25075019), a flavonoid found in food and red wine, can increase NKC activity.
* [Quercetin](https://www.ncbi.nlm.nih.gov/pubmed/19449452), a flavonoid in onions and fruits may improve NKC and T cell function.
* [Histone deacetylase inhibitors (HDACi) including suberoylanilide hydroxamic acid and valproic acid](https://www.ncbi.nlm.nih.gov/pubmed/17349632/) impair NKC function, and should be avoided.

T10927C (T;T)

**<function meter level="wildtype">**

**<variant and population data>**

**# Normal Function**

The CHRNE protein creates fully functional neural pathways and muscles.

**# What should I do about this?**

No therapies are medically indicated at the moment.

T10927C (T;C)

**<function meter level="wildtype">**

**<variant and population data>**

**# Heterozygous**

The CHRNE protein creates fully functional neural pathways and muscles.

**# What should I do about this?**

No therapies are medically indicated at the moment.

T10927C (C;C)

**<function meter level="high risk">**

**<variant and population data>**

**# High Risk**

Congenital myasthenic syndromes are a group of rare disorders that affects the proteins at the neuromuscular junctions and may cause [abnormalities in the CHRME receptors](https://www.ncbi.nlm.nih.gov/pubmed/16156017). It causes easy tiredness, muscle weakness, [scoliosis, and repetitive-compound muscle action.](https://www.ncbi.nlm.nih.gov/pubmed/27779167) Symptoms may worsen during [pregnancy](https://www.ncbi.nlm.nih.gov/pubmed/23108489). Other variants associated with myasthenic syndrome include the [AA epsilon1267delG deletion variant](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=244116), [1033-1G>C: splice acceptor variant](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=410057), [971delT deletion variant](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=33387), and [130dupG duplication variant](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=244117).

**# What should I do about this?**

Consult [a neurologist](https://www.ncbi.nlm.nih.gov/pubmed/23108489) during and after pregnancy. It afflicted with slow channel syndrome, consider adding [salbutamol in addition to fluoxetine](https://www.ncbi.nlm.nih.gov/pubmed/23281026). [Galantamine](http://www.uniprot.org/uniprot/Q04844) is also used in treatment.

**<symptoms>**

**<references>**

**<creator comment section>**

**What should I do about this?**

<symptoms fatigue >

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