

## What is Marinesco-Sjogren Syndrome (MSS)?

MSS is a very rare genetic disorder characterized by:

- cataracts (clouding of the lenses of the eyes, generally in early childhood)
- ataxia (balance and coordination problems)
- muscle weakness
- mental retardation (generally mild to moderate)
- very short stature
- dysarthria (slow, slurred speech)
- hypogonadism (decreased function of the ovary/testis).

## How is MSS diagnosed?

MSS is diagnosed based on the symptoms above. The child tends to be “floppy” and early childhood motor milestones are delayed. Cataracts develop, sometimes quickly. Growth is very slow. Most children have delays in academic, language, social, and self-help skills.

## How is MSS inherited?

MSS is an autosomal recessive disorder. Parents of an affected child have a 25% chance that any other child will have MSS. It is equally common in boys and girls. Mutations of the Sil1 gene are responsible for about 50% of all cases of MSS. There are milder variants with normal stature and minimal muscle weakness. The Sil1 gene plays a role in normal protein folding.

## How frequently does MSS occur?

MSS occurs in all ethnic groups. It is very rare except in a few genetically isolated groups. About 200 cases have been reported worldwide in the medical literature. There may be many more undiagnosed cases.

## What about treatment?

There is currently no treatment for MSS, but physical, occupational, and speech therapy are beneficial, along with special education services. Cataract surgery is required when vision becomes impaired. Hormone

replacement is needed if hypogonadism is present. Some children wear braces on their feet to improve ankle and foot alignment.

## What is the prognosis?

Most patients with MSS use a walker or crutches. Adults generally require some degree of support for daily living. Life expectancy is near-normal, but muscle weakness may increase with age.

## What are the research goals?

Researchers are working to identify all genes responsible for MSS (allowing accurate diagnosis, carrier testing, and prenatal diagnosis), understand the physiological basis of MSS, and eventually develop a treatment and cure for the disease. Identification of as many cases of MSS as possible is an important step towards these goals.

To learn more about MSS, meet other families, receive our newsletter, join our on-line discussion group, or contact researchers studying MSS,

visit the MSS Website at:

[www.marinesco-sjogren.org](http://www.marinesco-sjogren.org)

email us at:

[mss@marinesco-sjogren.org](mailto:mss@marinesco-sjogren.org)

or write to us at:

MSS Support Group  
1640 Crystal View Circle  
Newbury Park, CA 91320



### **Marinesco-Sjogren Syndrome Support Group**

- Providing support to families affected by Marinesco-Sjogren Syndrome (MSS)
- Reaching out to expand our network of MSS families
- Promoting research into diagnosis, treatment, and a cure for MSS

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