

What Is CAPS?

This booklet is your guide for all you need to know about CAPS. In it, you will learn what CAPS is, who gets CAPS, and what causes CAPS, as well as facts about CAPS symptoms and treatments.

If you think you or a family member has CAPS, here are some questions that may be used to help talk to a doctor:

- Do you get frequent rashes, fevers, or headaches?
- Do you feel a strong reaction to cold temperature?
- Do you always feel like you have the flu?
- Does your body ache constantly?
- Have you felt the same aches since you were a child?
- Do your aches seem to be getting worse?
- Have any of your family members or relatives ever suffered similar symptoms?
- Do any of your family members or relatives have CAPS?

Bring this booklet to the doctor's office to help remember what to address.

It is important that you speak with your doctor to see if you are suffering from CAPS. Your doctor can provide treatment to help relieve symptoms and, in some cases, eliminate them.

Facts About CAPS

Your Guide to Learning About Cryopyrin-Associated Periodic Syndrome (CAPS):

- **FCAS**—Familial Cold Autoinflammatory Syndrome
- **MWS**—Muckle-Wells Syndrome
- **NOMID/CINCA**—Neonatal-Onset Multisystem Inflammatory Disease, also known as Chronic Infantile Neurological Cutaneous Articular Syndrome

What Is CAPS?

CAPS stands for Cryopyrin-Associated Periodic Syndrome. It is the name given to a group of 3 different diseases.

These 3 diseases are:

- **FCAS**, which stands for Familial Cold Autoinflammatory Syndrome
- **MWS**, which stands for Muckle-Wells Syndrome
- **NOMID/CINCA**, which stands for Neonatal-Onset Multisystem Inflammatory Disease, also known as Chronic Infantile Neurological Cutaneous Articular Syndrome



CAPS is a group of rare inflammatory diseases.¹ Fewer than 1000 cases of CAPS have been reported in the world,² but more than 5500 people may actually have CAPS and not know it.³

The symptoms of all 3 types of CAPS are a result of the body's reaction to inflammation.⁴ The body responds to inflammation with soreness and swelling.

Some people may not know that they have CAPS,⁴ but their symptoms will make them aware that something is wrong. If you think you may have CAPS after reading this brochure, you should see a doctor.

If the doctor diagnoses you with CAPS, he will provide treatment to address the inflammation and any symptoms that may follow.

Please see page 13 for the meanings of key words that you may not know in this booklet.

The Facts on FCAS



FCAS stands for Familial Cold Autoinflammatory Syndrome. It can also be called FCAIS or FCU, which stands for Familial Cold Urticaria.

Symptoms of FCAS are less severe than the other 2 types of CAPS,⁴ but they can still seriously limit the decisions you make in your day-to-day life about what you can or cannot do.⁵ These symptoms include:

- A rash that keeps coming back⁴
- Headache⁴
- Fever⁴
- Chills⁴
- Feeling weak or tired⁴
- Sore or red eyes⁶
- Pain in the joints⁴
- Pain in the muscles⁴

Noticing Symptoms

People with FCAS usually start having symptoms from birth. Occasionally, symptoms may start later in childhood.⁴

Symptoms of FCAS can begin after a person is exposed to cold temperature.⁴ For example, being in an air-conditioned room, eating cold foods, or swimming in cool water can bring on symptoms.

A rash will usually start 1 to 2 hours after being exposed to the cold. Other symptoms may start 4 to 6 hours after being exposed to the cold.⁷

There may be some days when symptoms are not felt, but they always come back. These symptoms may get worse throughout the day. Most symptoms last up to 24 hours, although some can last longer.⁴

Some people may not know that they have FCAS.⁴ If you think you do, you should see your doctor.

It is important to be diagnosed and get treatment from your doctor. Treatment can help relieve symptoms and, in some cases, may eliminate them.



Rash in FCAS

FCAS Can Disrupt Your Life

In most cases, people that have FCAS know that something is wrong. It is clear to them from their symptoms that they need to see a doctor and find out what is going on with their health.

The symptoms of FCAS can seriously impact your day-to-day activities. Thirty people with FCAS were surveyed to see how FCAS had disrupted their everyday lives before they were treated.⁵

Here Is What They Said⁵:

- Almost all people with FCAS have had to give up most of their time spent outside



- Many people with FCAS said they have had to cancel plans with friends and family



- Many people with FCAS said that, even if they could work, it was still hard



- More than a third of the people with FCAS have had to leave their jobs



The symptoms of FCAS can strongly affect daily life. There are a number of treatments, however, that can help relieve symptoms and, in some cases, may eliminate them.

Please see page 10 for some of these treatments.

The Facts on MWS



MWS stands for Muckle-Wells Syndrome. MWS was first recognised in a family in the early 1960s by 2 doctors named Thomas James Muckle and Michael Vernon Wells.⁸ Since then, doctors have learned much more about MWS.

The symptoms of MWS listed below in **orange** are similar to FCAS symptoms, but they occur more often and may last longer.⁴ The symptoms in **green** are additional symptoms that someone with MWS may have.

- A rash that keeps coming back⁴
- Fever⁴
- Feeling weak or tired⁴
- Pain in the joints⁴
- High protein levels in the kidneys⁶
- Headache⁴
- Chills⁴
- Sore or red eyes⁶
- Pain in the muscles⁴
- Gradual loss of hearing⁴

Noticing Symptoms

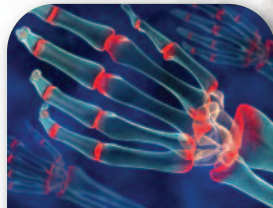
Like FCAS, people with MWS usually start having symptoms from birth. Occasionally, symptoms may start later in childhood.⁴

Symptoms can occur without any obvious reason. Only a few things have been linked to starting or making symptoms of MWS worse. For example, symptoms can begin after a person is exposed to cold temperature, feels stressed, or when the body is tired.⁴

There may be some days when symptoms are not felt, but they always come back. These symptoms may start anytime after being set off. Most symptoms last 1 to 2 days, although some can last longer.⁴

Some people may not know that they have MWS,⁴ but their symptoms will make them aware that something is wrong. If you think you have MWS, you should see your doctor.

It is important to be diagnosed and get treatment from your doctor. Treatment can help relieve symptoms and, in some cases, may eliminate them.



Pain in the joints in MWS

The Facts on NOMID



NOMID stands for Neonatal-Onset Multisystem Inflammatory Disease. NOMID can also be called CINCA, which stands for Chronic Infantile Neurological Cutaneous Articular Syndrome.

NOMID/CINCA is the most severe type of CAPS.⁴ The symptoms of NOMID/CINCA listed below in **green** are similar to MWS symptoms, but they are felt all the time.⁴ The symptoms in **blue** are additional symptoms that someone with NOMID/CINCA may have.

- A rash that keeps coming back⁴
- Fever⁴
- Feeling weak or tired⁴
- Pain in the joints⁴
- High protein levels in the kidneys⁶
- Swelling of the knees⁶
- Headache⁴
- Chills⁴
- Sore or red eyes⁶
- Pain in the muscles⁴
- Gradual loss of hearing⁴
- Gradual loss of eyesight⁴
- Mental difficulties⁴

Noticing Symptoms

People with NOMID/CINCA almost always start having symptoms from birth.⁴ A doctor may diagnose NOMID/CINCA quickly in a newborn because symptoms are often more noticeable than in FCAS or MWS.^{9,10}

NOMID/CINCA can be disabling¹ and, without treatment, some cases may result in death.¹¹ Treatment can help symptoms to some degree, but they never completely go away.⁴

It is clear to parents of children with NOMID/CINCA how much they suffer. From the moment they are born, it is a constant battle to keep NOMID/CINCA from getting worse.

For patients with all types of CAPS, but especially for those who need support for NOMID/CINCA, there are organisations that can help.

Please see page 12 for some informative Web sites.



Swelling of the knees in NOMID/CINCA

Why Haven't I Heard of CAPS?

CAPS is rare.¹ Fewer than 1000 cases of CAPS have been reported in the world,² but more than 5500 people may actually have CAPS and not know it.³ Because CAPS is so rare, most doctors will not have seen a patient with CAPS.¹⁰

Milder symptoms, such as headache and fever, can appear to be due to other things.^{4,6} It can take some time before a doctor begins to think about CAPS because it is not something he or she sees every day.⁴

Even those who have milder symptoms of CAPS may think their symptoms are due to the flu. So, they may not visit the doctor to ask about these symptoms.⁴

If you think you have CAPS, you should talk to your doctor. Bring this booklet with you so that you can remember the symptoms you want to talk to your doctor about.

It is important to be diagnosed and get treatment from your doctor. Treatment can help relieve symptoms and, in some cases, may eliminate them.

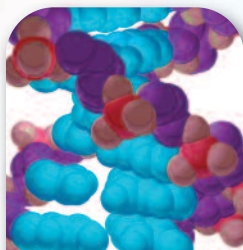
How Does Someone Get CAPS?

CAPS is usually passed on from one parent to the child through the genes.⁴ So, it is likely that someone who has CAPS got it from a parent.

Just because someone in your family has CAPS doesn't mean that you do too.⁶

Some people can have CAPS without anyone else in their family having it. However, for most, CAPS can be traced back in their family for many generations.⁶

If someone in your family has CAPS, you should talk to your doctor.



How Do I Know if I Have CAPS?

Any of the symptoms of the 3 types of CAPS can range from extremely uncomfortable to disabling in severe cases.^{4,5} Even if they are mild and don't seem to always be there, it is important to bring these symptoms to a doctor's attention.

Think about the answers to these questions:

- Do you get frequent rashes, fevers, or headaches?
- Do you feel a strong reaction to cold temperature?
- Do you always feel like you have the flu?
- Does your body ache constantly?
- Have you felt the same aches since you were a child?
- Do your aches seem to be getting worse?
- Have any of your family members or relatives ever suffered similar symptoms?
- Do any of your family members or relatives have CAPS?

Talk to your doctor about your answers to these questions. Details about your symptoms and your family history will help the doctor make the correct diagnosis.

Bring this booklet along with you to help you remember what you want to talk about.

It is important to be diagnosed and get treatment from your doctor. Treatment can help relieve symptoms and, in some cases, may eliminate them.

If you do have CAPS, your doctor will arrange to treat the symptoms with one of the medications available.

New Treatments for CAPS

Two Treatments Are Now Approved Specifically for CAPS

There are only two treatment options approved to treat the symptoms of CAPS. These treatments are injections and are available by prescription only.

One of the treatments is an injection that is given once a week. It is approved for use in FCAS and MWS in CAPS sufferers aged 12 years and older.

The other treatment is an injection that is given once every 8 weeks. This treatment is approved for use in FCAS, MWS, and NOMID/CINCA in CAPS sufferers aged 4 and older.

If you have been diagnosed with CAPS, one of these treatments may be right for you. Ask your doctor about these approved options.

What Else Can I Do?

If you have FCAS or MWS, your doctor may also suggest these actions to ease your symptoms⁴:

- Dress warmly with extra layers of clothing
- Drink hot beverages throughout the day
- Soak in a warm bath
- Cut back on heavy exercise and other strenuous activities
- Avoid things that cause you stress on a regular basis

In some serious cases, these changes may not help. CAPS sufferers who get cold easily and find their symptoms becoming worse may consider living somewhere where the weather is warmer.⁴ This can help relieve symptoms.



Where Else Can I Find Information?

There are support groups and organisations online that offer information about CAPS. The main goal is to help people understand these diseases and help improve the lives of people with CAPS.

Visit any of the sites below to learn more:

- CAPS Family Network—<http://www.capsfamilynetwork.com>
- The NOMID Alliance—<http://www.nomidalliance.net>
- The Genetics Home Reference Guide—<http://ghr.nlm.nih.gov>
- The Portal for Rare Diseases and Orphan Drugs—<http://www.orpha.net>
- National Organization for Rare Disorders (NORD)—<http://www.rarediseases.org/>
- International Society of Systemic Auto-Inflammatory Diseases (ISSAID)—<http://fmf.igh.cnrs.fr/ISSAID/>
- Centre de Référence des Maladies Auto-Inflammatoires—<http://asso.orpha.net/CEREMAI/index.html>

By browsing the Internet, you may come across other Web sites that support CAPS and allow people with CAPS to share their experiences. However, your doctor's advice is the best advice to follow.

Key Words

Antihistamines: drugs commonly used to treat allergies.

Articular: having to do with the joints in your body.

Autoinflammatory: inflammation in the body that occurs on its own without infection or injury.

Chronic: constant or frequent symptoms that keep coming back.

Clinical trials: studies used to determine if a new drug is approved.

Cutaneous: having to do with the skin.

Diagnose: a doctor confirms that you have an illness.

Disease: an illness that is set apart from others by the symptoms you may feel. It may also be called a *syndrome*.

Genes: when an illness is passed on through the genes, it means you can get it from your family or relatives. For example, if your parents have brown hair, you will likely have brown hair. Likewise, if your parents have CAPS, you are likely to also have CAPS.

High protein levels: proteins are the building blocks for all the parts of your body. For example, they make up the cells that act as your body's natural defence. Small amounts of protein may normally be found in your kidneys. But high protein levels in your kidneys may suggest an infection and should be treated by your doctor at once.

Immunosuppressants/immunomodulators: drugs commonly used to reduce inflammation.

Infantile: having to do with a baby, usually before walking age.

Inflammation: the body's natural defence to protect itself against infection or injury. In CAPS patients, inflammation occurs without infection or injury.

Inflammatory disease: an illness that is caused by inflammation. CAPS is a specific type of inflammatory disease called an autoinflammatory disease. See *autoinflammatory* definition above.

Neonatal: having to do with a newborn baby.

Neurological: having to do with the brain, the spine, and the nerves in your body.

Orphan drug: medicine that treats a rare disease.

Placebo: a treatment that has no medicine but is given in place of an actual treatment, usually as part of a clinical trial.

Rare disease: an uncommon illness that affects very few people in the world.

Steroids: drugs commonly used to treat a range of conditions, from irritation of the skin to joint pain.

Symptoms: a description of the way you are feeling due to a specific illness.

Syndrome: a group of symptoms that describe a specific illness. It may also be called a *disease*.

Urticaria: skin rash caused by inflammation.

