

# Familial Cold Autoinflammatory Syndrome (FCAS): Fact Sheet

TOPICS:

What It Is

Causes and Symptoms

Living with FCAS

Diagnosis and Treatment

## What It Is

Familial Cold Autoinflammatory Syndrome (FCAS) is a subtype of Cryopyrin-Associated Periodic Syndromes (CAPS). Diagnosis usually occurs shortly after birth.<sup>1</sup>

## Causes and Symptoms

Symptoms of FCAS are similar to (but should not be confused with) acquired cold urticaria, a more common condition that is mediated by different mechanisms and seldom appears during infancy.<sup>1</sup> FCAS is generally caused by mutations in a gene called *NLRP3* (nucleotide-binding domain, leucine-rich family [NLR] pyrin domain containing 3). *NLRP3* mutations cause increased activity of cryopyrin, a protein that regulates inflammation. Increased cryopyrin activity causes overproduction of a protein called interleukin-1 beta (IL-1 $\beta$ ).<sup>2,3</sup>

Overproduction of IL-1 $\beta$ , in turn, causes recurrent symptoms of inflammation such as<sup>3</sup>

- Rash
- Fever/chills
- Joint pain
- Eye redness/pain
- Fatigue

Symptoms may be triggered by exposure to cooling temperatures. In most cases, a rash occurs within a few hours, followed by fever and joint pain. Most flares resolve within 24 hours if further exposure to cooling temperatures is avoided.<sup>1</sup>

## **Living with FCAS**

Patients may try to reduce their symptoms by controlling their environment and limiting their exposure to cold. They may wear multiple layers of clothing, keep warm temperatures in their homes, and stay indoors without air-conditioning.<sup>4</sup>

## **Diagnosis and Treatment**

Diagnosis of FCAS is determined through an evaluation of a patient's symptoms. Confirmation of the diagnosis is sometimes achieved through genetic testing and the identification of *NLRP3* mutations.<sup>5</sup> Not all patients, however, have a detectable genetic mutation, making accurate symptom evaluation critical.<sup>2</sup>

Some of the common criteria that distinguish FCAS from other hereditary periodic fevers include<sup>1</sup>

- Recurrent, intermittent episodes of fever and rash that primarily follow exposure to cooling temperatures
- Family history of the disease
- Symptoms usually appear shortly after birth
- Duration of most attacks <24 hours
- Presence of eye redness/pain associated with attacks

There are FDA-approved therapies to treat the symptoms of FCAS.<sup>6</sup>