



PATIENT FACT SHEET

Cryopyrin-Associated Autoinflammatory Syndromes (CAPS) (Juvenile)



CONDITION DESCRIPTION

Cryopyrin-associated autoinflammatory syndromes (CAPS) are three very rare diseases related to a defect in the cryopyrin protein. Different organ systems may be involved. Severity varies among patients. CAPS diseases usually start in very young children.

CAPS include Neonatal Onset Multisystem Inflammatory Disease (NOMID), Muckle-Wells Syndrome and familial

cold autoinflammatory syndrome. CAPS is caused by a gene mutation that may be inherited from only one parent. The gene mutation causes a defect in the cryopyrin protein, which helps control inflammation. Males and females from all ethnic groups may have CAPS. NOMID is the most severe and least severe CAPS disease.



SIGNS/ SYMPTOMS

NOMID causes fever and inflammation in multiple organs. It may affect newborns with a fever and hives-like rash that does not itch. Patients may have chronic meningitis, causing headache, blindness, hearing loss and neurologic problems. Children's eyes may bulge, and they may have vomiting episodes. By age 1, half of children with NOMID develop pain and swelling in large joints such as the knee. Growth delay may occur. Children with NOMID are often very short.

Signs of Muckle-Wells include episodic fever, chills, rash, red eyes, joint pain and severe headaches with vomiting. Episodes last from one to three days. Children may develop partial or total hearing loss by their teen years.

Children with familial cold autoinflammatory syndrome react to cold exposure with a hives-like rash, as well as fever, chills, nausea, severe thirst, red eyes, headaches and joint pain. Episodes may last for a day.



COMMON TREATMENTS

A pediatric rheumatologist can diagnose CAPS. Diagnosis is based on physical examination and genetic testing. Some patients may have normal genetic tests. Skin biopsy of a rash, eye exam, hearing tests, lumbar puncture to obtain fluid from the spine, and magnetic resonance imaging (MRI) of the brain or inner ears may be needed.

Medications that target interleukin-1 are very effective CAPS treatments. These are injections and include anakinra [Kineret], rilonacept [Arcalyst] and canakinumab

[Ilaris]. Treatment must continue for life. There is no cure for CAPS.

Early diagnosis and treatment may prevent damage to bone, brain, eyes or hearing that often cannot be repaired once it has occurred. Patients with severe headaches or brain involvement may need higher doses or more frequent injections.

Physical therapy and splints may help children with joint deformities. Surgery is occasionally needed. Children with hearing loss may need hearing aids.



CARE/ MANAGEMENT TIPS

Children whose CAPS is not treated may have severe episodes that lower quality of life. Children may miss school, and families may have psychological and financial stress. They may need psychological support to cope.

Children with familial cold autoinflammatory syndrome may need to live in mild climates and avoid places with very cold air conditioning. With new, effective

treatments, children may be able to control CAPS symptoms and lead near-normal lives. It is important to continue treatment for life as prescribed.

Lifelong treatment may also prevent amyloidosis, a possible complication. Amyloid proteins can deposit into organs like the kidneys, as well as the heart, intestines and skin. Loss of kidney function is possible. Children must take their treatment even when they are feeling better.