

Muckle-Wells Syndrome (MWS): Fact Sheet

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What It Is

MWS is one of the Cryopyrin-Associated Periodic Syndromes (CAPS) generally caused by mutations in the CIAS1/NLRP-3 gene. These syndromes are characterized by recurrent attacks of rash, fever/chills, joint pain, fatigue, and eye pain/redness. MWS symptoms are triggered by unknown random factors and possibly by stress, exercise, or cold.

In most cases, Muckle-Wells Syndrome (MWS) patients develop progressive hearing loss. In some MWS cases amyloidosis, a disease in which an abnormal accumulation of the protein, amyloid, occurs in a patient's tissues and organs, develops later in life.

Causes and Symptoms

MWS is generally caused by a mutation in a gene identified as the cold-induced auto-inflammatory syndrome 1 (CIAS1) gene, more recently named the Nod-Like Receptor Protein-3 (NLRP-3) gene. The CIAS1/NLRP-3 gene mutation is passed on in an autosomal dominant manner. The mutation also can occur spontaneously. The mutation in the CIAS1/NLRP-3 gene causes increased activity of cryopyrin, a protein that regulates inflammation in the body. The increased activity of cryopyrin results in an overproduction of a protein known as interleukin-1 β (IL-1 β), which leads to symptoms of inflammation such as fever and joint pain. Common symptoms of MWS, which are generally first experienced during early childhood or adolescence, include recurrent rash, fever/chills, joint pain, fatigue, eye pain/redness, progressive hearing loss, and amyloidosis. Symptoms can vary between individuals,

MWS symptoms are triggered by unknown random factors and possibly by stress, exercise, or cold. Episodes generally last between 24 to 48 hours.

Incidence and Prevalence

MWS is thought to be a very rare condition. Since MWS is a newly discovered condition, the actual incidence and prevalence of the disease is difficult to determine.

Living with MWS

The symptoms and complications related to MWS can sometimes be quite debilitating.

Diagnosis & Treatment

Diagnosis of MWS is determined through an evaluation of a patient's symptoms. Confirmation of the diagnosis is sometimes, but not always achieved through genetic testing and the identification of a CIAS1/NLRP-3 gene mutation.

To correct the hearing loss that often occurs, hearing aids may be used. There are no medications currently approved for the treatment of MWS by the U.S. Food and Drug Administration.