

# Yao Syndrome

Subjects: **Genetics & Heredity**

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Yao syndrome (formerly called *NOD2*-associated autoinflammatory disease) is a disorder involving episodes of fever and abnormal inflammation affecting many parts of the body, particularly the skin, joints, and gastrointestinal system.

genetic conditions

## 1. Introduction

Inflammation is a normal immune system response to injury and foreign invaders (such as bacteria). In people with Yao syndrome, part of the immune system called the innate immune response is turned on (activated) abnormally, which causes fevers and inflammation-related damage to tissues and organs. Based on this process, Yao syndrome is classified as an autoinflammatory disease. Autoinflammatory diseases are distinct from autoimmune diseases; these two groups of diseases involve abnormalities in different parts of the immune system.

The episodes of fever and inflammation associated with Yao syndrome can last for several days and occur weeks to months apart. During these episodes, most affected individuals develop reddened, inflamed areas on the skin called erythematous patches or plaques. This reddening occurs most commonly on the face, chest, and back but can also affect the arms and legs. Episodes of joint pain and inflammation similar to arthritis are common, particularly in the legs, as is swelling of the ankles and feet. Inflammation also affects the gastrointestinal system, causing attacks of abdominal pain, bloating, and cramping with diarrhea in more than half of affected individuals. Dry eyes and dry mouth (described as "sicca-like" symptoms, which refers to dryness) are reported in about half of people with this disease. Other potential signs and symptoms of Yao syndrome include mouth sores, chest pain, and enlargement of various glands.

Yao syndrome is usually diagnosed in adulthood. It is a long-lasting (chronic) disease, and episodes can recur for many years.

## 2. Frequency

Yao syndrome has an estimated prevalence of 1 in 10,000 to 1 in 100,000 people worldwide. Studies suggest that it is among the most common systemic (affecting the whole body) autoinflammatory diseases in adults. For unknown reasons, Yao syndrome appears to affect women more frequently than men.

## 3. Causes

The causes of Yao syndrome are complex. This condition likely results from a combination of genetic and environmental factors, many of which are unknown.

Certain variations in the *NOD2* gene increase the risk of developing Yao syndrome. The *NOD2* protein plays several essential roles in the immune system's response to foreign invaders, including inflammatory reactions. Studies suggest that most people with Yao syndrome have at least one variation in the *NOD2* gene, and some have two or more. It is unclear what effect these variations have on the amount or function of the *NOD2* protein, or how they might contribute to abnormal inflammation in people with Yao syndrome. Researchers suspect that environmental factors such as infections may also play a role in triggering the disease in people with genetic variants that increase their risk.

### 3.1 The gene associated with Yao syndrome

- *NOD2*

## 4. Inheritance

Because Yao syndrome appears to be a complex disease without a single genetic cause, it does not have a straightforward pattern of inheritance. A small percentage of affected individuals have a family history of the disease. Many people who have one or more of the *NOD2* gene variants associated with Yao syndrome never develop the disease.

## 5. Other Names for This Condition

- NAID
- *NOD2*-associated AID
- *NOD2*-associated autoinflammatory disease
- YAOS

## References

1. Estephan M, Yao Q, Springer J. Case of *NOD2*-Associated Autoinflammatory Disease Successfully Treated With Sulfasalazine. *J Clin Rheumatol*. 2017 Jan;23(1):58-59. doi: 10.1097/RHU.0000000000000468.
2. Yao Q, Lacbawan F, Li J. Adult autoinflammatory disease frequency and our diagnostic experience in an adult autoinflammatory clinic. *Semin Arthritis Rheum*. 2016 Apr;45(5):633-7. doi: 10.1016/j.semarthrit.2015.10.012.

3. Yao Q, Myles J, Shen B, McDonald C. NOD2-associated autoinflammatory disease: an exploratory study of its pathogenesis. *Rheumatology (Oxford)*. 2014May;53(5):958-60. doi: 10.1093/rheumatology/ket384.
4. Yao Q, Shen B. A Systematic Analysis of Treatment and Outcomes of NOD2-Associated Autoinflammatory Disease. *Am J Med*. 2017Mar;130(3):365.e13-365.e18. doi: 10.1016/j.amjmed.2016.09.028.
5. Yao Q, Shen M, McDonald C, Lacbawan F, Moran R, Shen B. NOD2-associated autoinflammatory disease: a large cohort study. *Rheumatology (Oxford)*. 2015Oct;54(10):1904-12. doi: 10.1093/rheumatology/kev207.
6. Yao Q, Su LC, Tomecki KJ, Zhou L, Jayakar B, Shen B. Dermatitis as a characteristic phenotype of a new autoinflammatory disease associated with NOD2 mutations. *J Am Acad Dermatol*. 2013 Apr;68(4):624-631. doi:10.1016/j.jaad.2012.09.025.
7. Yao Q, Zhou L, Cusumano P, Bose N, Piliang M, Jayakar B, Su LC, Shen B. A new category of autoinflammatory disease associated with NOD2 gene mutations. *Arthritis Res Ther*. 2011;13(5):R148. doi: 10.1186/ar3462.

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