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## **Blau's Disease / Juvenile Sarcoidosis**

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### **1. WHAT IS BLAU'S DISEASE/JUVENILE SARCOIDOSIS**

#### **1.1 What is it?**

Blau syndrome is a genetic disease. Patients suffer from a combination of skin rash, arthritis and uveitis. Other organs may be affected and intermittent fever may be present as well. Blau syndrome is the term used for familial forms of the disease but sporadic forms can occur as well and are known as Early Onset Sarcoidosis (EOS).

#### **1.2 How common is it?**

The frequency is unknown. It is a very rare disease that affects patients in early childhood (mostly before 5 years of age) and becomes worse if left untreated. Since the discovery of the associated gene, it is being diagnosed more frequently, which will allow for a better estimate of prevalence and natural history.

#### **1.3 What are the causes of the disease?**

Blau syndrome is a genetic disease. The responsible gene is called NOD2 (synonymous with CARD15), which encodes a protein with a role in the immune-inflammatory response. If this gene carries a mutation, as in Blau syndrome, the protein does not function properly and patients experience chronic inflammation with granuloma formation in various tissues and organs of the body. Granulomas are characteristic long-lived clusters of inflammatory cells that are associated with inflammation and may disrupt the normal structure and functioning of various tissues and organs.

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#### **1.4 Is it inherited?**

It is inherited as an autosomal dominant disease (which means that it is not linked to gender and that at least one parent must show symptoms of the disease). This type of transmission means that to have Blau syndrome, an individual needs only one mutated gene, either from the mother or the father. In EOS, the sporadic form of the disease, the mutation emerges in the patient him/herself and both parents are healthy. If a patient carries the gene, he or she will suffer from the disease. If one parent has Blau syndrome, there is a 50% chance that his/her child will suffer from it.

#### **1.5 Why does my child have this disease? Can it be prevented?**

The child has the disease because he/she carries genes that cause Blau syndrome. Currently, the disease cannot be prevented but the symptoms can be treated.

#### **1.6 Is it infectious?**

No, it is not.

#### **1.7 What are the main symptoms?**

The main symptoms of the disease are a clinical triad of arthritis, dermatitis and uveitis. The initial symptoms comprise a typical exanthema, with tiny round lesions that vary in colour from pale pink to tan or intense erythema. Over the course of years, the rash waxes and wanes. Arthritis is the most common manifestation, starting in the first decade of life. There is joint swelling with preserved mobility at onset. With time, limitation of movement, deformities and erosions may develop. Uveitis (inflammation of the iris) is the most threatening manifestation, since it is often associated with complications (cataract, increased intraocular pressure) and may cause decreased vision if left untreated.

In addition, granulomatous inflammation may affect a wide spectrum of other organs, causing other symptoms, as well such as decreased lung or kidney functioning, increased blood pressure or recurrent fever.

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### **1.8 Is the disease the same in every child?**

It is not the same in every child. Moreover, the type and severity of symptoms may change as the child grows older. The disease progresses if left untreated and the symptoms will evolve accordingly.