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NLRP-12 Related Recurrent Fever

Version of 2016

1. WHAT IS NLRP-12-RELATED RECURRENT FEVER

1.1 What is it?

NLRP-12-related recurrent fever is a genetic disease. The responsible gene is called NLRP12 (or NALP12), which has a role in inflammatory signalling pathways. Patients suffer from recurrent bouts of fever accompanied by a variety of symptoms such as headache, joint paint or joint swelling and skin rash. Symptoms are probably triggered by cold exposure. Untreated, the disease could be very debilitating but it is not life-threatening.

1.2 How common is it?

The disease is very rare. Currently, fewer than 10 patients have been identified worldwide.

1.3 What are the causes of the disease?

NLRP-12 related recurrent fever is a genetic disease. The responsible gene is called NLRP12 (or NALP12). The genetically modified gene is responsible for a disturbance of the inflammatory response of the body. The exact mechanism of this disturbance is still under investigation.

1.4 Is it inherited?

NLRP-12 related recurrent fever is inherited as an autosomal dominant disease. This means that to have NLRP-12 related recurrent fever, an individual needs to have one affected parent. Sometimes, there is no

other member of the family suffering of recurrent fever: either the gene has been damaged at the child's conception (known as de novo mutation) or the parent who carries the mutation may not exhibit the clinical symptoms or exhibits only a very mild form of the disease (variable penetrance).

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

The child has the disease because he/she inherited the disease from one parent who carries a NLRP-12 gene mutation, unless a de novo mutation has occurred. The person who carries the mutation may or may not exhibit the clinical symptoms of NLRP-12-related recurrent fever. The disease cannot currently be prevented.

1.6 Is it infectious?

NLRP-12 related recurrent fever is not an infectious disease. Only genetically-affected subjects can develop the disease.

1.7 What are the main symptoms?

The main symptom is fever. Fever lasts about 5-10 days and recurs at irregular intervals (weeks to months). The bouts of fever are accompanied by a variety of symptoms. These may include headache, joint pain and joint swelling, urticarial rash and myalgia. Fever bouts are probably triggered by cold exposure. In one family only, sensorineural hearing loss was observed.

1.8 Is the disease the same in every child?

The disease is not the same in every child; the disease varies between a mild and a more severe form. Moreover, the type, duration and severity of attacks may be different each time, even in the same child.

1.9 Is the disease in children different from the disease in adults?

As patients grow up, the fever attacks seem to become fewer and milder. However, some disease activity will remain in most if not all

affected individuals.