www.printo.it/pediatric-rheumatology/GB/intro

TNF Receptor Associated Periodic Syndrome (TRAPS)

Version of 2016

1. WHAT IS TRAPS

1.1 What is it?

TRAPS is an inflammatory disease characterised by recurrent attacks of high spiking fevers, usually of two to three weeks duration. The fever is typically accompanied by gastrointestinal disturbances (abdominal pain, vomiting, diarrhoea), painful red skin rash, muscle pain and swelling around the eyes. Impaired kidney function can be observed in the late phase of the disease. It is possible to observe similar cases in the same family.

1.2 How common is it?

TRAPS is thought to be a rare disease but the true prevalence is currently unknown. It affects males and females equally and the onset is usually during childhood, although patients with an adult onset have been described.
The first cases were reported in patients of Irish-Scottish ancestry; however, the disease has also been identified in other populations: French, Italians, Sephardic and Ashkenazi Jews, Armenians, Arabs and Kabylians from Maghreb.
The seasons and climate have not been demonstrated to influence the course of the disease.

1.3 What are the causes of the disease?

TRAPS is due to an inherited anomaly of a protein (Tumour Necrosis Factor Receptor I [TNFRI]), which leads to an increase of the patient's normal acute inflammatory response. TNFRI is one of the cellular receptors specific for a potent inflammatory circulating molecule known as tumour necrosis factor (TNF). The direct link between the alteration of the TNFRI protein and the severe recurrent inflammatory state observed in TRAPS has not been completely identified yet. Infection, injury or psychological stress may trigger the attacks.

1.4 Is it inherited?

TRAPS is inherited as an autosomal dominant disease. This form of inheritance means the disease is transmitted by one of the parents who has the disease and carries an abnormal copy of TNFRI gene. All individuals have 2 copies of all genes; hence, the risk of an affected parent transmitting the mutated copy of the gene TNFRI to each child is 50%. De novo (new) mutation may also occur; in such cases, neither parent has the disease and neither carries a mutation of the TNFRI gene but disruption of TNFRI gene appears at the child's conception. In this case, the risk of another child developing TNFRI is random.

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

TRAPS is an inherited disease. A person who carries the mutation may or may not exhibit the clinical symptoms of TRAPS. The disease cannot currently be prevented.

1.6 Is it infectious?

TRAPS is not an infectious disease. Only genetically-affected individuals develop the disease.

1.7 What are the main symptoms?

The main symptoms are recurrent attacks of fever typically lasting two or three weeks but sometimes of shorter or longer duration. These episodes are associated with chills and intense muscle pain involving the trunk and the upper limbs. The typical rash is red and painful, corresponding with underlying inflammation of the skin and muscle

area.
Most patients experience a sensation of deep cramping muscle pain at the onset of attacks that gradually increases in intensity and begins to migrate to other parts of the limbs, followed by the appearance of a rash. Diffuse abdominal pain with nausea and vomiting are common. Inflammation of the membrane covering the front of the eye (the conjunctiva) or swelling around the eyes is characteristic of TRAPS, although this symptom can be observed in other diseases. Chest pain due to inflammation of the pleura (the membrane surrounding the lungs) or of the pericardium (the membrane surrounding a joint) is also reported.
Some patients, especially in adulthood, have a fluctuating and sub-chronic disease course, characterised by flares of abdominal pain, joint and muscle aches, ocular manifestations with or without fever and a persistent elevation of laboratory parameters of inflammation.
Amyloidosis is the most severe long-term complication of TRAPS, occurring in 14% of patients. Amyloidosis is due to tissue deposition of a circulating molecule produced during inflammation, called serum amyloid A. Renal deposition of amyloid A leads to the loss of a large amount of proteins in the urine and progresses to renal failure.

1.8 Is the disease the same in every child?

TRAPS presentation varies from one patient to another in terms of the duration of each attack and the duration of symptom-free periods. The combination of the main symptoms is also variable. These differences may be explained in part by genetic factors.