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Majeed Syndrome

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1. WHAT IS MAJEED

1.1 What is it?

Majeed syndrome is a rare genetic disease. Affected children suffer from Chronic Recurrent Multifocal Osteomyelitis (CRMO), Congenital Dyserythropoietic Anaemia (CDA) and inflammatory dermatosis.

1.2 How common is it?

The disease is very rare and described only families of Middle East origin (Jordan, Turkey). The actual prevalence is estimated at less than 1/1,000,000 children.

1.3 What are the causes of the disease?

The disease is caused by mutations in the LPIN2 gene on chromosome 18p that codes for a protein called lipin-2. Researchers believe that this protein may play a role in the processing of fats (lipid metabolism). However, no lipid abnormalities have been found with Majeed syndrome.
Lipin-2 also may be involved in controlling inflammation and in cell division.
Mutations in the LPIN2 gene alter the structure and function of lipin-2. It is unclear how these genetic changes lead to bone disease, anaemia and inflammation of the skin in people with Majeed syndrome.

1.4 Is it inherited?

It is inherited as an autosomal recessive disease (which means that it is not linked to gender and that neither parent necessarily has symptoms of the disease). This type of transmission means that to have Majeed Syndrome, an individual needs two mutated genes, one from the mother and the other from the father. Hence, both parents are carriers (a carrier has only one mutated copy but not the disease) and not patients. Although carriers typically do not show signs and symptoms of the condition, some parents of children with Majeed syndrome have had an inflammatory skin disorder called psoriasis. Parents who have a child with Majeed syndrome have a 25% risk that another child will have the same disease. Antenatal diagnosis is possible.

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

The child has the disease because it was born with the mutated genes that cause Majeed Syndrome.

1.6 Is it infectious?

No, it is not.

1.7 What are the main symptoms?

Majeed Syndrome is characterised by chronic recurrent multifocal osteomyelitis (CRMO), congenital dyserythropoietic anaemia (CDA) and inflammatory dermatosis. The CRMO associated with this syndrome can be differentiated from isolated CRMO by an earlier age at onset (in infancy), more frequent episodes, shorter and less frequent remissions and the fact that it is probably life-long, leading to retarded growth and/or joint contractures. CDA is characterised by peripheral and bone marrow microcytosis. It can be variable in severity, ranging from mild, unnoticeable anaemia to a blood transfusion-dependent form. The inflammatory dermatosis is usually Sweet syndrome but can also be pustulosis.

1.8 What are the possible complications?

CRMO can lead to complications such as slow growth and the development of joint deformities called contractures, which restrict the

movement of certain joints; the anaemia may result in symptoms including tiredness (fatigue), weakness, pale skin, and shortness of breath. Complications of congenital dyserythropoietic anaemia can range from mild to severe.

1.9 Is the disease the same in every child?

Due to the extreme rarity of this condition, little is known about the variability of the clinical manifestations. In any case, the severity of symptoms may vary among different children leading to a milder or more severe clinical picture.

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

Little is known about the natural history of the disease. In any case, adult patients present more disabilities related to the development of complications.