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Papa Syndrome (Piogenic Arthritis, Pioderma, Gancrenosum and Acne)

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1. WHAT IS PAPA

1.1 What is it?

The acronym PAPA stands for Pyogenic Arthritis, Pyoderma gangrenosum and Acne. It is a genetically determined disease. A triad of symptoms that includes recurrent arthritis, a type of skin ulcers known as pyoderma gangrenosum and a type of acne known as cystic acne characterises the syndrome.

1.2 How common is it?

PAPA syndrome appears to be very rare. Very few cases have been described. However, the frequency of the disease is not known exactly and may be underestimated. PAPA affects male and females equally. Usually, the disease appears during childhood.

1.3 What are the causes of the disease?

PAPA syndrome is a genetic disease caused by mutations in a gene called PSTPIP1. The mutations change the function of the protein for which the gene codes; this protein plays a role in the regulation of the inflammatory response.

1.4 Is it inherited?

PAPA syndrome is inherited as an autosomal dominant disease. This

means that it is not linked to gender. It also means that one parent shows at least some symptoms of the disease and usually more than one affected individual is be observed in a single family, with affected individuals in each generation. When someone with PAPA syndrome is planning to have children, there is a 50% chance of having a child with PAPA syndrome.

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

The child has inherited the disease from one of his/her parents who carries a mutation in the PSTPIP1 gene. The parent who carries the mutation may or may not exhibit all symptoms of the disease. The disease cannot be prevented but the symptoms can be treated.

1.6 Is it infectious?

PAPA syndrome is not infectious.

1.7 What are the main symptoms?

The most common symptoms of the disease are arthritis, pyoderma gangrenosum and cystic acne. Rarely are all three present in the same patient at the same time. Arthritis occurs usually early in childhood (the first episode occurs between 1 and 10 years of age); it usually involves one joint at a time. The affected joint becomes swollen, painful and red. The clinical appearance resembles septic arthritis (arthritis caused by presence of bacteria in the joint). Arthritis of PAPA syndrome may cause damage to articular cartilage and periarticular bone. The large ulcerative skin lesions, known as pyoderma gangrenosum, usually have a later onset and often involve the legs. Cystic acne usually appears during adolescence and may persist into adulthood, involving the face and trunk. Symptoms are often precipitated by minor injury to the skin or joint.

1.8 Is the disease the same in every child?

The disease is not the same in every child. An individual carrying a mutation in the gene may not exhibit all the symptoms of the disease or may exhibit only very mild symptoms (variable penetrance). Moreover,

the symptoms may change, usually improving, as a child grows older.