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

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2. DIAGNOSIS AND TREATMENT

2.1 How is it diagnosed?

It is possible to consider PAPA syndrome in a child with repeated episodes of painful inflammatory arthritis that clinically resemble septic arthritis and do not respond to antibiotic treatment. Arthritis and skin manifestations may not appear at the same time and may not be present in all patients. A detailed evaluation of the family history should also be performed; since the disease is autosomal dominant, other family members are likely to exhibit at least some symptoms of the disease. The diagnosis can only be made by genetic analysis to ascertain the presence of mutations in the PSTPIP1 gene.

2.2 What is the importance of tests?

Blood tests: erythrocyte sedimentation rate (ESR), C-reactive protein (CRP) and blood cell counts are usually abnormal during the episodes of arthritis; these tests are used to demonstrate the presence of inflammation. Their abnormalities are not specific for the diagnosis of PAPA syndrome.
Joint fluid analysis: during episodes of arthritis, joint puncture to obtain joint fluid (known as synovial fluid) is usually performed. Synovial fluid from patients with PAPA syndrome is purulent (yellow and dense) and contains an elevated number of neutrophils, a type of white blood cell. This feature is similar to septic arthritis but bacterial cultures are negative. Genetic test: the only test that unambiguously confirms the

diagnosis of PAPA syndrome is a genetic test that shows the presence of a mutation in the PSTPIP1 gene. This test is performed on a small amount of blood.

2.3 Can it be treated or cured?

Since it is a genetic disease, PAPA syndrome cannot be cured. However, it can be treated with drugs that control inflammation in joints, preventing joint damage. The same is true for skin lesions, although their response to treatment is slow.

2.4 What are the treatments?

The treatment of PAPA syndrome is different depending on the dominant manifestation. Arthritis episodes usually respond rather promptly to oral or intra-articular corticosteroids. Occasionally, their efficacy may not be satisfactory and arthritis may also recur very often, necessitating long-term corticosteroids that may cause to side effects. Pyoderma gangrenosum shows some response to oral corticosteroids and is also usually treated with local (cream) immunosuppressant and anti-inflammatory drugs. The response is slow and the lesions may be painful. Recently, in single cases, treatment with new biologic drugs that inhibit IL-1 or TNF have been reported to be efficacious for both pyoderma and for treating and preventing the recurrences of arthritis. Because of the rarity of the disease, no controlled studies are available.

2.5 What are the side effects of drug therapy?

Corticosteroid treatment is associated with weight gain, swelling of the face and mood changes. Long-term treatment with these drugs may cause suppression of growth and osteoporosis.

2.6 How long should treatment last?

Treatment is usually aimed at controlling recurrences of arthritis or of skin manifestations and usually it is not administered continuously.

2.7 What about unconventional or complementary therapies?

There are no published reports of effective complementary therapies.

2.8 How long will the disease last?

Affected individuals usually get better as they grow older and disease manifestations may disappear. However, this does not occur in all patients.

2.9 What is the long-term prognosis (predicted outcome and course) of the disease?

Symptoms get milder with age. However, since PAPA syndrome is a very rare disease, the long-term prognosis is not known.