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Cryopyrin Associated Periodic Syndromes (CAPS) (CINCA/Muckle Wells/FCAS)

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2. DIAGNOSIS AND THERAPY

2.1 How is it diagnosed?

Diagnosis of CAPS is based on clinical symptoms before being genetically confirmed. Distinction between FCAS and MWS or MWS and CINCA/NOMID might be difficult because of overlapping symptoms. Diagnosis is based on clinical symptoms and medical history of the patient. Ophthalmologic evaluation (in particular fundoscopy), CSF examination (lumbar puncture) and radiological evaluation are helpful in distinguishing contiguous diseases.

2.2 Can it be treated or cured?

CAPS cannot be cured since they are genetic diseases. However, thanks to substantial advances in the understanding of these disorders, new promising drugs are now available to treat CAPS and are under investigation for their long-term effect.

2.3 What are the treatments?

Recent work on the genetics and physiopathology of CAPS shows that IL-1β, a powerful cytokine (protein) of inflammation, is overproduced in these conditions and plays a major role in the onset of the disease. Currently, a number of drugs that inhibit IL-1β (IL-1 blockers) are in various stages of development. The first drug used in treating these conditions was anakinra. It was shown to be rapidly efficient in

controlling inflammation, rash, fever, pain and fatigue in all CAPS. This treatment also effectively improves neurological involvement. In some conditions, it may improve deafness and control amyloidosis. Unfortunately, this drug does not seem to be effective on overgrowth arthropathy. Doses required depend on disease severity. Treatment must be started early in life, before chronic inflammation causes irreversible organ damage such as deafness or amyloidosis. It requires daily subcutaneous injection. Local reactions at the injection site are frequently reported but may resolve with time. Rilonacept is another anti-IL-1 drug approved by the FDA (Food and Drug Administration in USA) for patients older than 11 years who suffer from FCAS or MWS. Weekly subcutaneous injections are required. Canakinumab is another anti-IL-1 drug recently approved by the FDA and European Medicines Agency (EMA) for CAPS patients older than 2 years. In MWS patients, this drug has been recently shown to effectively control the inflammatory manifestations with a subcutaneous injection every 4 to 8 weeks. Due to the genetic nature of the disease, it is conceivable that the pharmacological blockade of IL-1 should be maintained for long periods, if not life-long.

2.4 How long will the disease last?

CAPS are life-long disorders.

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

The long-term prognosis of FCAS is good but quality of life can be affected by recurrent episodes of fever. In MWS syndrome, the long-term prognosis may be affected by amyloidosis and impaired renal function. Deafness is also a significant long-term complication. Children with CINCA may have growth disturbances during the course of the disease. In CINCA/NOMID, the long-term prognosis depends on the severity of neurological, neurosensorial and joint involvements. Hypertrophic arthropathies may impose severe disabilities. Premature death is possible in severely affected patients. Treatment with IL-1 blockers has greatly improved the outcome of CAPS.