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CRYOPYRIN-ASSOCIATED PERIODIC SYNDROME

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Cryopyrin-associated periodic syndrome (CAPS) also known as NLRP3-associated auto inflammatory disease is a collection of conditions including familial cold auto inflammatory syndrome type 1 (FCAS1), Muckle-Wells syndrome (MWS), and neonatal-onset multisystem inflammatory disorder (NOMID). It is caused by a mutation in the gene NLRP3 which codes for the cryopyrin protein, which is a protein involved in immunity. We present two clinical cases on CAPS.

Case 1 Boy S., 15 yo, has initially presented at the age of three yo with complaints of prolonged high grade fever, rash and leg pain. Two months prior the boy was seen by hematologist due to elevated inflammatory markers. Past medical history was remarkable for recurrent episodes of fever and rush since the age of 7 months and one episode of inability to stand on feet followed by several days of ankle joint being swollen and skin warm on palpation at the age of 1 yo. Systemic-onset Juvenile Idiopathic Arthritis (JIA) was diagnosed and treatment with systemic corticosteroids initiated. Response was good but on the attempts to reduce the dose recurrence of fever and rash was observed. The patient's symptoms were controlled with prolonged corticosteroid's treatment. Short stature was revealed at the age of 5 yo and sensorineural deafness at the age of 8. The boy was assessed by geneticist and the genetic testing revealed mutation in NLRP3 gene. Since 2021 the patient has been treated with Canakinumab which allowed to get rid of steroids. No exacerbations have been noted since the treatment has been started.

Case 2. 1 year old girl was followed by primary physician for persisting elevation of ESR noted at the age of 1 year during routine check-up. Later the girl developed high grade fever and rash and was admitted to an infectious disease hospital. During her stay, she continued to have a high fever, a rash, arthritis of ankle joints and inflammatory changes in blood tests. Multisystem inflammatory syndrome was suspected and treatment with corticosteroid pulse therapy and immunoglobulin I/V infusions initiated. Initial treatment was effective and fever subsided, patient has been discharged home. But few weeks after pulse therapy the girl continued to have fever and inflammatory changes in blood tests. Juvenile idiopathic arthritis with a systemic onset (hematological syndrome, lymphadenopathy, hepatosplenomegaly, pneumonitis, anemia, exanthema, articular syndrome and fever), activity grade 3 was diagnosed. Methylprednisolone 1 mg/kg and methotrexate 10 mg/m2 were prescribed, but there was no significant clinical improvement noticed. Slow growth velocity and severe Cushing syndrome were observed. Following this, the girl was sent for genetic assessment, which revealed the mutation "NLRP 12c.2374G^A" (FCAS). Treatment with Canakinumab is initiated but no results yet are seen.

In conclusion, these two clinical cases are perfect examples as to how CAPS can easily be misdiagnosed as JIA. They share common symptoms such as recurrent fevers, joint pain, deforming joint disease and rashes. It also has to be noted that genetic testing plays a vital role when diagnosis CAPS. However upon treatment with canakinumab each patient's prognosis improved significantly.