SBR 2019
36º CONGRESSO BRASILEIRO DE REUMATOLOGIA
CENTRO DE EVENTOS DO CEARÁ
04 A 07 DE SETEMBRO
FOR TALEZA 2019
PROMOÇÃO
70 anos Sociedade Brasileira de Reumatologia
REALIZAÇÃO
SCR SOCIEDADE CEARENSE DE REUMATOLOGIA

THE USE OF ANTI-IL1 (CANAKINUMAB) IN A CHILD WITH MEVALONIC ACIDURIA: A CASE REPORT

Jéssica Aparecida Fernandes (UFMG, Belo Horizonte, MG, Brasil), Ihan Bruno Lopes Rabelo (UFMG, Belo Horizonte, MG, Brasil), Ana Luiza Garcia Cunha (Fundação Hospitalar de Minas Gerais, Belo Horizonte, MG, Brasil), Ericka Viana Machado Carellos (Fundação Hospitalar de Minas Gerais, Belo Horizonte, MG, Brasil), Fernanda Gontijo Minafra Silveira Santos (UFMG, Belo Horizonte, MG, Brasil), Luciana Araújo Oliveira Cunha (UFMG, Belo Horizonte, MG, Brasil), Flávia Patrícia Sena Teixeira Santos (UFMG, Belo Horizonte, MG, Brasil), Anna Carolina Faria Moreira Gomes Tavares (UFMG, Belo Horizonte, MG, Brasil)

BACKGROUND

Mevalonate kinase deficiency is a very rare autosomal recessive inheritance that occurs due to the MVK gene mutation located on the long arm of chromosome 12. This mutation leads to a deficiency of the enzyme mevalonate kinase, involved in the synthesis of isoprenoids, and manifests as recurrent fever and elevation of acute phase reactants, in addition to lymphadenomegaly and hepatosplenomegaly, abdominal pain, vomiting, skin lesions, oral aphtous ulcers and arthralgia or arthritis. There are different phenotypes, hyperimmunoglobulinemia D and periodic fever syndrome, the milder one, and mevalonic aciduria, the more severe one.

CASE REPORT

Patient, 11 months old, male, with accentuated delay in neuropsychomotor development, wide fontanelles, ocular deviation down and nystagmus on the left and severe malnutrition. He was born in the preterm delivery, at home, and from birth presented with anemia and hepatosplenomegaly. She evolved with multiple hospitalizations, always with fever, pallor, food refusal, prostration and respiratory discomfort, as well as tachycardia, tachypnea, elevation of inflammation tests and leukocytosis with neutrophilia. Clinical presentation was always treated as sepsis, but with negative blood cultures. During the intercrisis periods, he maintained food refusal and respiratory discomfort. At 5 months, he evolved with respiratory failure, fever, worsening of hepatosplenomegaly and pancytopenia. Laboratory tests demonstrated hypofibrinogenemia, hyperferritinemia, hypertriglyceridemia and increased transaminases, which led to the suspicion of hemophagocytic syndrome. Treatment with dexamethasone 10mg/m2/SC was initiated, with excellent clinical response. Myelogram was inconclusive. The subsequent clinical worsening coincided with attempts to reduce corticotherapy. Subsequently it evolves with polyarthritis (hands and right knee). Retrospective analysis of the case led to suspicion of autoinflammatory disease, and the positive dosage of mevalonic acid in urine confirmed the diagnosis of mevalonic aciduria. Treatment with canaquinumab 2mg/kg was started every 4 weeks. After the first dose, there was cessation of fever, improvement of both food acceptance and respiratory discomfort. The patient was discharged from hospital 2 weeks after the second dose. Waiting for genetic panel results.

CONCLUSION

Autoinflammatory diseases bring daily diagnostic challenges to clinical practice, both because they mimic infectious conditions and because of the difficult access to confirmatory tests. For this case, the anti-IL1 drug is considered the best therapy, although there may be no complete response. Despite the off-label use in children under 2 years of age and under 7.5 kg, it was chosen due to the severity of the condition and the scarce therapeutic options, obtaining an excellent response so far.