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POLYARTERITIS NODOSA OR DEFICIENCY OF ADENOSINE DEAMINASE 2?: CASE REPORT.

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BACKGROUND

Necrotizing vasculitis is a group of diseases characterized by inflammation of the vessels associated with necrosis and your main example is Polyarteritis Nodosa (PAN). PAN is characterized as a vasculitis of medium vessels manifested by systemic symptoms (fever, myalgias, arthralgia, weight loss) and specific organ involvement (livedo reticular, nodules, stroke, peripheral neuropathy, proteinuria, hematuria, gastrointestinal involvement). Another group of disorders associated with necrotizing vasculitis are monogenic autoinflammatory diseases which Deficiency of Adenosine Deaminase 2 (DADA-2) has a PAN-Like clinical presentation with genetic etiology (autosomal recessive mutation in the CERC1 gene). Considering the great similarity between the two entities and the need for early recognition for the establishment of correct therapy, we will describe two cases initially diagnosed and conducted as PAN and later identified absence of ADA-2 enzyme activity.

CASE REPORT

A 5-year-old male patient presented episodes of recurrent fever, myalgia, hepatosplenomegaly and lymph node enlargement. Laboratory tests showed negative autoantibodies. Myelogram with mild desieritropoese. Echocardiogram without abnormalities. Abdominal ultrasound with hepatomegaly and moderate splenomegaly. Induction treatment was instituted monthly with cyclophosphamide and maintenance with azathioprine and prednisone. He has been asymptomatic and lost follow-up for 10 years.

He returned to follow-up after new episodes of fever and myalgia with increased inflammatory evidence and encephalic MR angiography with signs of vasculitis. During this same period, his brother at 8-year-old presented fever, myalgia and elevated inflammatory activity, therefore was extensively investigated and demonstrated central nervous system vasculitis in brain MRI angiography.

There was no satisfactory response to treatment. The enzymatic activity of ADA-2 has been performed and showed a total absence of activity in both patients.

CONCLUSION

Due the great clinical similarity, it is necessary to suspect of DADA-2 in PAN associated with a similar family history, early stroke, recalcitrant PAN and early onset of PAN-like disease. According to the literature, platelet count can be used to differentiate the diseases mentioned. It is increased in PAN and normal in DADA-2. Imaging tests, as well as biopsies, present the same aspects in the two pathologies, and are not used to differentiate them. Early diagnosis is important because DADA-2 does not respond effectively to traditional immunosuppressive therapy, but has a satisfactory response due the use of anti-TNF.