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ERDHEIM-CHESTER DISEASE PRESENTING AS RETROPERITONEAL FIBROSIS

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BACKGROUND

Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytosis that is characterized by the accumulation of foamy histiocytes in the retroperitoneum, long bones, and large vessel areas. It may be a life-threatening condition, especially in cases of heart or central nervous system (CNS) involvement.

Almost 60% of ECD patients carry a BRAFV600E-activating mutation of the RAS-RAF-MEK-extracellular signal-regulated kinase signalling pathway.

CASE REPORT

Male, 53 years, develop back pain and reduction of urinary output, search for a urologist assistance who detected an abnormal kidney function triggered by a ureteral obstruction in the context of a retroperitoneal fibrosis. He successfully opted for a ureteral stenting and referred the patient to our rheumatology centre for diagnosis purpose.

In first evaluation patient had already bradycardia, pericardial effusion, diffuse pain and hypothyroidism.

The clinical investigation emphasized IGG4 related disease, among other causes of retroperitoneal fibrosis.

The biopsys of pericardium and retroperitoneal were inconclusive.

Bone scan was positive for osteosclerotic lesions and guided a bone biopsy which was suggestive of ECD, with typical accumulation of foamy histiocytes and supported by immunochemistry (table 1).

The evaluation of the BRAFV600E mutation was negative.

Treatment with prednisone in immunosuppressive doses failed to improve symptoms and was initiated interferon infusion with resolution of pain and a noteworthy benefit in quality of life.

CONCLUSION

The clinical presentation of patients with ECD varies depending on the sites of involvement. Most patients with ECD will have osseous involvement at the time of diagnosis and the vast majority will also have at least one non-osseous site of involvement.

In that case, a patient with abnormal kidney function was evaluated by urologist, who detected ureteral obstruction caused by a striking retroperitoneal fibrosis. By that time patient was also afflicted with hypothyroidism, bradycardia, pericardial effusion and diffuse pain.

This scenario was extremely challenging for a definite diagnosis. Albeit, multisystemic diseases frequently demands early treatment even though we lack an accurate diagnosis. ECD for instance, is a rare and highly specific illness in terms of pathophysiology and treatment, so we had to be certain, in order to provide the best treatment available. Immunohistochemistry was remarkable and our patient lack the BRAFV600e mutation.

The treatment with interferon stabilized the progression of the disease and improved symptoms. Although there is no evidence of benefit in survival rate.