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RESEARCH ARTICLE

BONE MARROW OXALOSIS, A RARE CAUSE OF PANCYTOPENIA: A CASE REPORT

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Abstract

Primary hyperoxaluria is a rare congenital metabolic disease characterised by an overproduction of oxalate, secondary to a hepatic enzyme deficiency, alanine glyoxylate aminotransferase (AGT). Calcium oxalate crystals are deposited in many tissues. Involvement of the bone marrow is relatively rare. We report the case of a young 29-year-old patient with a history of recurrent renal lithiasis, progressing to end-stage chronic renal failure at the haemodialysis stage, admitted to the medical ward for investigation of mucocutaneous pallor and anaemia resistant to erythropoietin. Clinical and laboratory examination revealed an oedematous-ascitic syndrome and pancytopenia with normocytic normochromic anaemia. Imaging revealed moderate ascites and homogeneous splenomegaly. The myelogram was inconclusive. An osteomedullary biopsy was performed and showed deposits of oxalate crystals with bone marrow fibrosis associated with foreign body macrophagic granulomas. These crystals were grouped in a rosette and birefringent in polarised light. Primary hyperoxaluria is a rare disease. Involvement of the bone marrow indicates the advanced stage of the disease, hence the importance of anatomopathological diagnosis. Hepato-renal transplantation remains the only effective treatment for this disease.

Introduction:-

Primary hyperoxaluria is a rare congenital metabolic disorder characterized by the overproduction of oxalate, secondary to a hepatic enzyme deficiency, alanine glyoxylate aminotransferase (AGT) **[1].** Its clinical presentation includes nephrocalcinosis and renal failure.

Deposition of excess oxalate occurs in bone and in all organs and tissues, but bone marrow involvement is relatively rare **[1].** It is characterized by cytopenias, leukoerythroblastosis, and hepatosplenomegaly **[2],** as well as birefringent calcium oxalate crystals on polarised microscopy and granulomatous structures in the bone marrow **[3].**

Hepato-renal transplantation remains the only effective treatment for this condition **[4].**

Case Observation:-

We report the case of a 29-year-old patient with a history of recurrent renal lithiasis, who progressed to end-stage chronic renal failure at the hemodialysis stage. He was admitted to the internal medicine unit for investigation of mucocutaneous pallor and anemia resistant to erythropoietin.

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Clinical examination revealed an oedematous ascitic syndrome.

The laboratory work-up showed pancytopenia with normochromic normocytic anemia, with a hemoglobin level of 5.8 g/dl, a white blood cell count of 1.2×10^9/L, and a platelet count of 98×10^9.

Radiological findings included moderate ascites and homogeneous splenomegaly.

The myelogram was inconclusive.
An osteomedullary biopsy was performed.

Microscopic examination of the bone marrow biopsy showed extensive deposits of numerous calcium oxalate crystals with bone marrow fibrosis associated with macrophagic foreign body granulomas **[Figure 1].** These crystals were grouped in a rosette and were birefringent in polarised light.
[Figure 2].

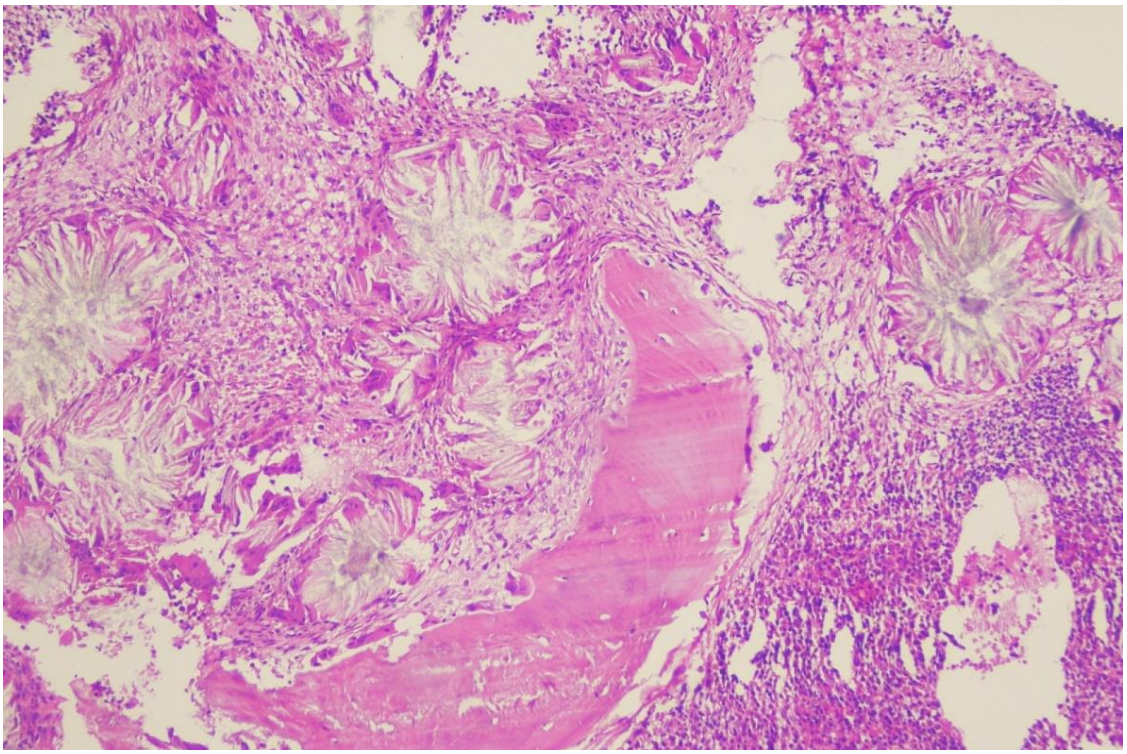

Figure 1:- Medullary tissue with crystal deposits and medullary fibrosis associated with foreign body macrophagic granulomas. X10.

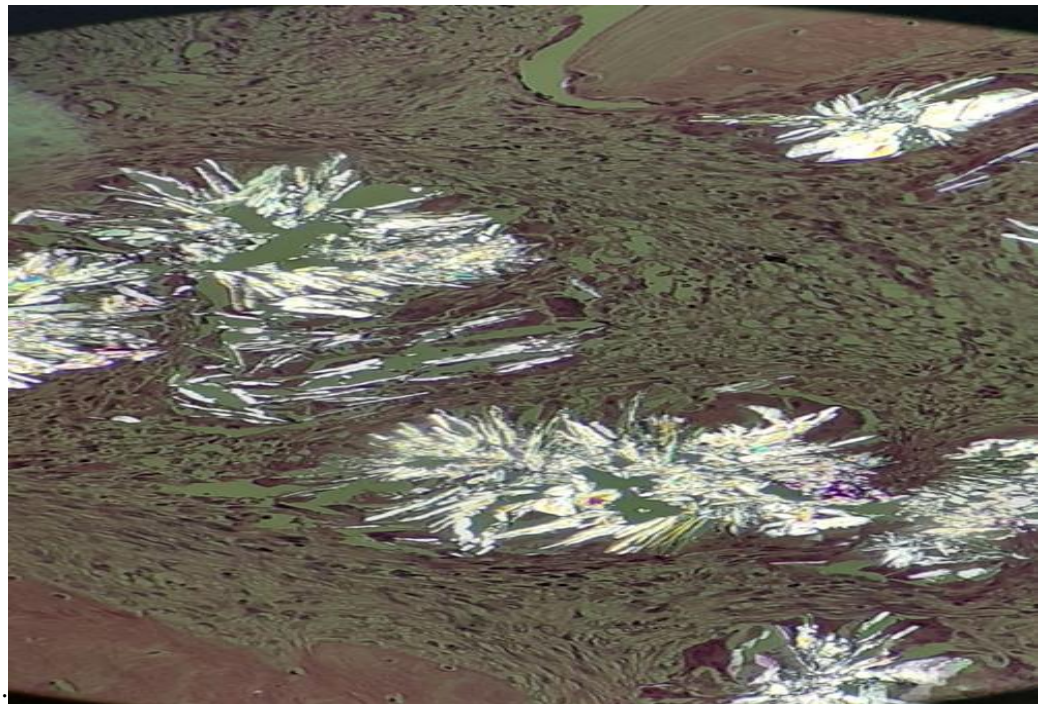

Figure 2:- déposits of birefringent crystals in polarised light.

Discussion:-

Primary hyperoxaluria (PH) is an extremely rare autosomal recessive disorder.

Its incidence is estimated at 1 in 120,000 cases per year.

It generally occurs in childhood or adolescence, with some cases diagnosed in the sixth decade of life **[5].**

Hyperoxaluria is said to be primary when it is caused by the overproduction of oxalate, due to an enzyme deficiency in the liver, alanine glyoxylate aminotransferase (AGT), and it may also be secondary due to increased intestinal absorption or overconsumption of oxalate-rich foods **[6].** Overfeeding was not reported in our patient.

Oxalose is a product that cannot be metabolized. It, therefore, accumulates in the body through excretion into the systemic circulation.

An early stage of the disease is defined by the recurrent formation of renal calculi, nephrocalcinosis, which alters the function of the kidneys and causes a reduction in urinary excretion, further increasing the deposition of oxalate in the systemic circulation and favoring systemic deposition of oxalate and progression to renal failure, as was the case in our patient **[6-7].**

Excess oxalate deposition occurs in bone and in all organs and tissues except the liver. Major organs include the retina, arteries, peripheral nervous system, myocardium, thyroid, skin, spinal cord, and medulla **[8,9].**

The clinical presentation depends on the location of the crystal deposits.

Medullary oxalosis is rare. It is characterized by the appearance of cytopenias; sometimes pancytopenias and organomegalia such as hepatosplenomegaly **[2].**

Without treatment, the prognosis for patients is very poor and can be fatal.

Combined liver and kidney transplantation is necessary for recovery **[4].**

Conclusion:-

In conclusion, primary hyperoxaluria is a rare metabolic disease that leads to end-stage renal failure. Involvement of bone marrow tissue by oxalate crystal deposits is rare and reflects the late stage of the disease. Hepato-renal transplantation is the treatment in this case.

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