**MEDULLARY OXALOSE: About a rare case**

**ABSTRACT :**

We report a case of medullary oxalosis in a patient with terminal chronic renal failure whose hematology laboratory performed a diagnostic work-up after pancytopenia was found. A 32-year-old patient receiving chronic hemodialysis who has had end-stage chronic renal failure for six years also has underlying cardiopathy. The patient's medical history began one month before admission, when significant asthenia, an indeterminate weight loss, and a left hypochondrial enlargement that hampered breathing first appeared. Primary hyperoxaluria can be effectively treated by a combined liver and kidney transplant, which replaces the damaged enzyme and creates a new excretion pathway for oxalate. Although systemic oxalosis can be stabilized or even reversed with effective treatment, tissue crystal clearance is sluggish; Therefore, in order to avoid irreparable organ damage, early diagnosis and treatment are essential. New forms of treatment have recently emerged, including RNA interference therapy. Prenatal diagnosis and genetic counselling have been made possible by our growing understanding of the AGXT gene and genetic advancements, raising the prospect of a revolutionary genetic engineering treatment soon.

**Keywords: bone marrow, pancytopenia, medullary oxalosis, and genetic engineering**

**INTRODUCTION :**

Medullary oxalosis is an autosomal recessive inherited metabolic disorder resulting from a deficiency of the hepatic enzyme alanine-glyoxylate aminotransferase (AGT) (1). This condition represents a form of systemic oxalosis characterized by the deposition of oxalate within the bone marrow (BM). Although rare, it can present with pancytopenia at advanced stages, accompanied by visceral involvement that may pose a significant risk to patient survival.

We present a case of medullary oxalosis in a patient with terminal chronic renal failure, whose diagnostic work-up was conducted in the Hematology laboratory following the identification of pancytopenia.

**CASE PRESENTATION :**

A 32-year-old patient with a history of end-stage chronic renal failure for 6 years, undergoing chronic hemodialysis, also presents with underlying cardiopathy. The patient's illness history dates back to one month prior to admission, marked by the onset of a left hypochondrial swelling that impeded respiration, severe asthenia, and unquantified weight loss. Clinical examination revealed splenomegaly extending 7 cm below the costal margin, without hepatomegaly. The hemogram showed pancytopenia with severe anemia (hemoglobin at 6.5 g/dL, normochromic normocytic), neutropenia (1285/mm³), and thrombocytopenia (40,000/mm³). The bone marrow aspirate was hypocellular, prompting a complementary bone marrow biopsy (BMB) that confirmed the presence of multiple oxalate crystal deposits within the marrow, accompanied by a foreign body giant cell reaction and no evidence of malignancy (Figure 1(A-C)). Therapeutically, the management included addressing transfusion needs with per-dialysis transfusions.

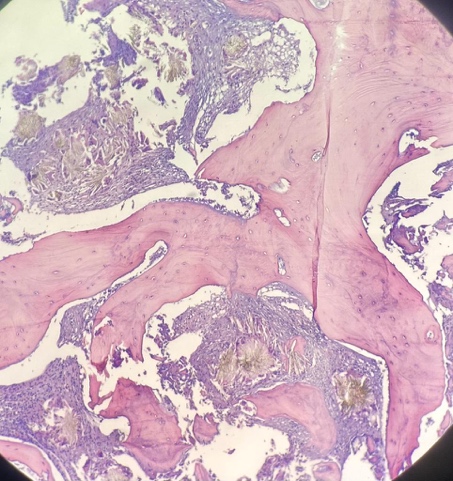
**DISCUSSION:**

With an approximate yearly incidence of less than one case per million people, medullary oxalosis is an extremely uncommon clinical condition. In Morocco, there has only been one instance reported thus far (2). There are few cases of bone marrow oxalosis in the literature. This condition is frequently linked to leukoerythroblastic responses, resistance to erythropoietin therapy, and cytopenias of various severity. One characteristic that is seen in the development of primary hyperoxaluria is the accumulation of calcium oxalate crystals in the bone marrow. However, side effects such extramedullary hematopoiesis and bone marrow failure are still rare. In clinical practice, medullary oxalosis is strongly suggested by the triad of pancytopenia, splenomegaly, and a dry tap on bone marrow aspiration.

In order to address the underlying enzyme deficiency and create a functioning channel for oxalate excretion, coupled liver and kidney transplantation is the last treatment for primary hyperoxaluria. Although systemic oxalosis can be stopped or even partially reversed with effective treatment, accumulated tissue oxalate crystals take time to remove. Therefore, preventing permanent end-organ damage requires early diagnosis and intervention. The landscape of treatment is changing due to recent developments in therapeutic possibilities, such as RNA interference therapy. These innovative treatments seek to lessen the severity of the illness and may lessen the need for liver transplantation in the future, which would diminish the morbidity that goes along with it (3).

**CONCLUSION :**

Pancytopenia associated with medullary oxalosis typically does not respond to stimulating agents and may necessitate repeated transfusions, as observed in this case. Understanding the AGXT gene and advances in genetics have enabled prenatal diagnosis and genetic counseling, creating hope for a radical treatment through genetic engineering in the near future.

**A close-up of a pink and white cell

Description automatically generatedA pink and purple cells under a microscope

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**FIGURE 1(A-C) :** Bone marrow biopsy : Multiple deposits of oxalate crystals within the marrow, accompanied by a foreign body giant cell reaction and no signs of malignancy.

**CONFLICTS OF INTEREST :**

No conflicts of interest

**Ethical Approval:**

As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

**CONSENT :**

The authors declare that they have obtained consent from the family of the patient discussed in the report.

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1.

2.

3.

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