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Bilateral Exophthalmos Revealing Acute Myeloid Leukemia in an Infant: Case Report

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Abstract **Case Report**

Acute myeloid leukemia (AML) is a malignancy characterized by the uncontrolled proliferation of immature hematopoietic cells, leading to the infiltration of various tissues. Ocular involvement in AML is rare and typically associated with a poor prognosis. A 1-year-old infant with no significant medical history presented with rapidly progressive bilateral proptosis, more pronounced on the left side, over a 15-day period. Ophthalmological examination revealed painful, inflammatory proptosis and stage 2 bilateral papilledema. CT scans indicated diffuse infiltration of the orbital and extraconical spaces, along with proptosis. Blood tests showed pancytopenia, and a myelogram confirmed the diagnosis of AML. Unfortunately, the child's condition deteriorated due to severe sepsis, leading to death following hospitalization in the pediatric ICU. Ocular involvement in acute leukemia is uncommon but may present as chloroma or granulocytic sarcoma, typically manifesting as rapidly progressive unilateral or bilateral exophthalmos. This can be mistaken for conditions such as rhabdomyosarcoma or infection. The presence of leukemia in the oculo-orbital region is a poor prognostic indicator, as it is considered a central nervous system disorder and should be managed accordingly. The prevalence of ocular involvement has significantly decreased with the advent of appropriate chemotherapy.

Keywords: exophtalmos; inflammatory proptosis; myeloid leukemia.

1) INTRODUCTION

Acute myeloid leukemia (AML) is an aggressive hematologic malignancy defined by the clonal proliferation of immature myeloid precursors, resulting in the progressive replacement of normal hematopoietic elements within the bone marrow and subsequent infiltration of peripheral tissues. The uncontrolled accumulation of these blasts leads to impaired hematopoiesis, manifesting clinically as anemia, thrombocytopenia, and recurrent infections.

Although leukemic infiltration of ocular structures is relatively uncommon compared to other systemic manifestations, its occurrence is of particular concern due to its strong association with unfavorable outcomes. Ocular involvement may compromise both visual prognosis, through direct infiltration or secondary complications, and overall survival, as it frequently reflects widespread disease or central nervous system extension. Consequently, the recognition of ocular manifestations in AML has important diagnostic, prognostic, and therapeutic implications, underscoring the necessity of comprehensive ophthalmologic evaluation in patients with suspected or confirmed leukemia

2) CASE REPORT

This 1-year-old infant, with no significant medical history, was referred to the ophthalmology emergency department for bilateral proptosis, more severe on the left side, which rapidly progressed over a period of 15 days.

The ophthalmological examination revealed bilateral, painful, inflammatory proptosis that could not be reduced, developing in a context of asthenia, pallor, and deterioration of the general condition (Fig. 1). The anterior segment examination was normal, and the fundus examination revealed stage 2 bilateral papilledema.

The remainder of the physical examination did not reveal a tumor syndrome. Orbital-cerebral CT scan demonstrated diffuse infiltration of the intra- and

extraconical space as well as the oculomotor muscles, associated with bilateral proptosis, more pronounced on the left side (Fig. 2).

Blood tests revealed pancytopenia, and a myelogram confirmed the diagnosis of acute myeloid leukemia. The child's course was marked by the onset of severe sepsis, leading to his death after hospitalization in the children's intensive care unit.

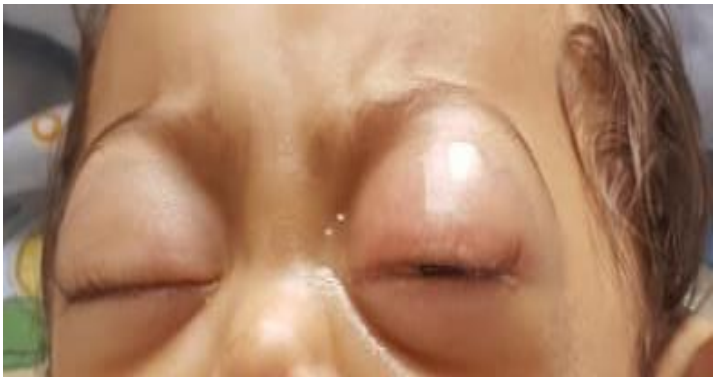

Figure 1: Bilateral exophthalmos, axillary inflammation more marked in the left eye

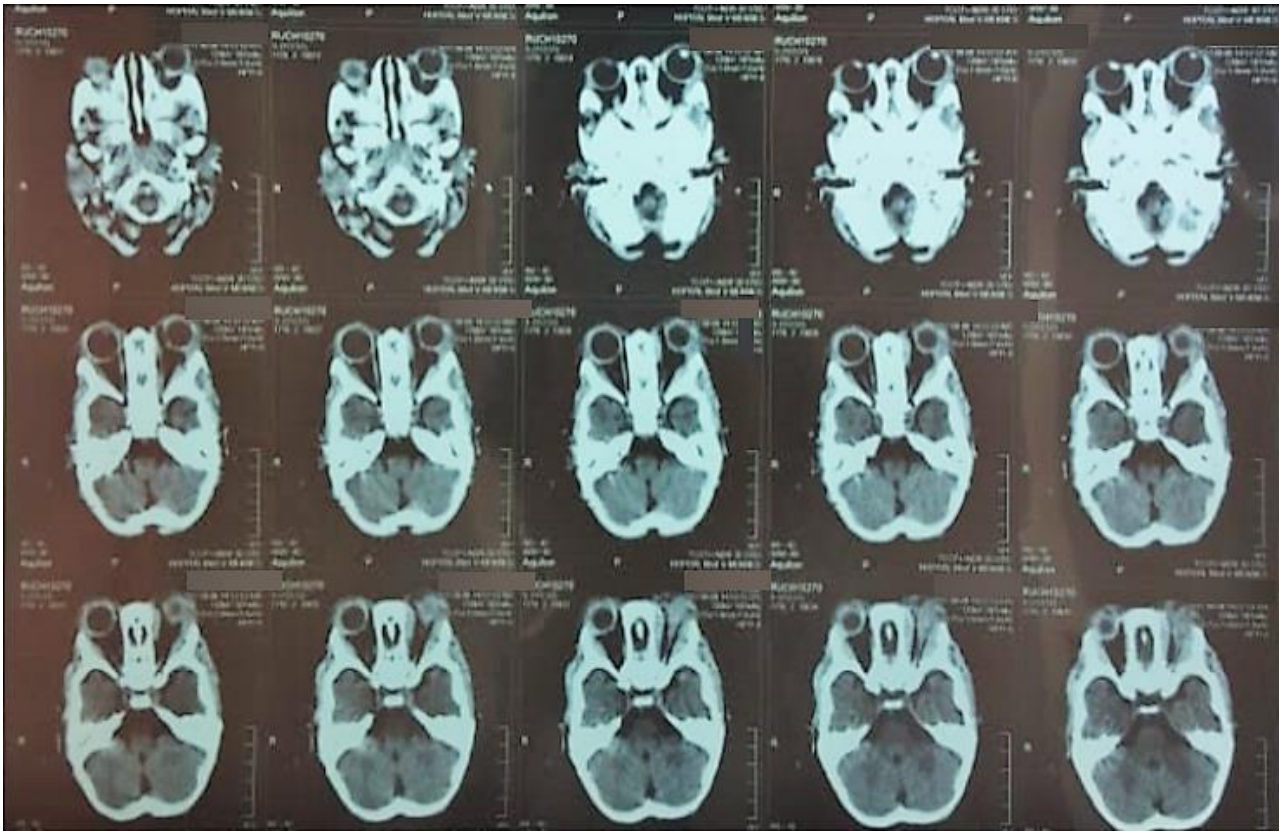

Figure 2: Orbital-cerebral computed tomography showing infiltration of the extra- and intra-conical space

3) DISCUSSION

The prevalence of ocular manifestations in acute leukemia shows considerable variability across different studies. However, recent advances in chemotherapeutic protocols have contributed to a significant reduction in such complications [1].

Orbital infiltration represents an uncommon but clinically significant manifestation. This entity, referred to as chloroma or granulocytic sarcoma, frequently constitutes the initial presentation of the disease. Clinically, it is typically characterized by a rapidly progressive, unilateral or bilateral inflammatory exophthalmos. Such presentation may closely mimic other orbital pathologies, including rhabdomyosarcoma, or masquerade as inflammatory and infectious orbital disorders [2].

Importantly, the occurrence of ocular or oculo-orbital involvement in leukemia is associated with an unfavorable prognosis [3-5]. This manifestation is

currently regarded as an extension of central nervous system (CNS) disease. Consequently, it necessitates management strategies analogous to those employed in CNS leukemia, emphasizing the need for prompt recognition and aggressive therapeutic intervention.

4) CONCLUSION

Orbital infiltration in the context of leukemia represents a particularly severe manifestation, associated with both a poor functional and vital prognosis. Its presence not only threatens visual integrity but also reflects advanced systemic disease, often correlating with unfavorable overall outcomes. Consequently, any ophthalmologist should maintain a high index of suspicion when confronted with a case of rapidly progressive unilateral or bilateral exophthalmos in a pediatric patient. Early recognition of this presentation is critical, as timely diagnosis and initiation of appropriate systemic therapy may significantly influence both survival and quality of life.

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