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Acute Myeloid Leukemia Presenting as Bilateral Proptosis and Bilateral Skull Metastatic Lesions

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Introduction: Acute myeloid leukemia (AML), constituting 30% of pediatric malignancies, is the most common childhood cancer. This paper explores the rare presentation of AML with extramedullary involvement, specifically bilateral proptosis, in a 15-year-old boy.

Case report: The patient initially presented with worsening shortness of breath, palpitations, extreme fatigue, and bruising. Examination revealed bilateral proptosis, watery discharge from the right eye, and petechial rash. Blood investigations revealed low hemoglobin, severe thrombocytopenia, and high WBC count. CT revealed bilateral orbital infiltrative soft tissue lesions. Leukemia fusion gene screening identified RUNX1-RUNX1T1. The patient was admitted, received transfusions, and started on antibiotics. Despite initial improvement, he later developed sepsis, septic shock, and severe pancytopenia, necessitating intensive care and specific AML M2 targeting therapy.

Conclusion: Bilateral proptosis in AML, termed myeloid sarcoma (MS), is rare but responsive to chemotherapy. Orbital MS has higher responsiveness and survival rates in pediatric cases. The case highlights the importance of identifying AML subtypes, like RUNX1-RUNX1T1 and CBFB-MYH11-positive AML, for tailored treatment strategies. This case underscores the challenges in diagnosing and treating pediatric AML with extramedullary involvement. Early recognition of AML subtypes is crucial for prognosis prediction and treatment tailoring.