Case Reports

Choroidal and Orbital Leukemic Infiltrate Mimicking Advanced Retinoblastoma

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INTRODUCTION

The ophthalmic manifestations of acute leukemia in children include iris infiltrates, hyphema, hypopyon, vitreous infiltrates, retinal or preretinal infiltrates, leukemic retinopathy, choroidal infiltrates, optic nerve infiltration, and orbital infiltration.\(^1\)\(^-\)\(^3\) Leukemic retinopathy usually denotes the fundus manifestations of anemia, thrombocytopenia, and hyperviscosity.\(^4\) The choroid is the most commonly affected ocular tissue, but choroidal involvement is often not clinically apparent.\(^1\) Leukemic infiltration of the orbit, called a leukemic choroma, occurs more commonly in acute leukemia.\(^1\)

The presenting signs of retinoblastoma include\(^6\) leukokoria, strabismus, poor vision in the affected eye(s), orbital inflammation and proptosis, red painful eye due to secondary glaucoma, and spontaneous hyphema, as well as other less common signs. Patients with advanced retinoblastoma can present with leukokoria, orbital cellulitis due to inflammation from tumor necrosis,\(^5\) and proptosis due to extraocular extension.

In this article, we present a patient with acute lymphoblastic leukemia who presented with leukokoria and proptosis due to an extensive intraocular and orbital tumor. We do not believe that this presentation of acute leukemia has been reported, although massive choroidal infiltration has been mentioned in one article.\(^6\) The purpose of this article is to add another entity to the differential diagnosis of leukokoria with proptosis, and to demonstrate how the correct diagnosis was established.

CASE REPORT

A previously healthy 6-month-old boy presented with a 3-week history of subcutaneous scalp nodules and a 2-day history of proptosis of the left eye. Ophthalmic evaluation showed good fixation with the right eye, but poor fixation with the left eye. Both pupils were reactive, and no afferent pupillary defect was present. External and anterior segment examination of the right eye was entirely normal. Examination of the left eye showed proptosis with inferior displacement of the globe, and upper lid edema (Fig 1). A pink, fleshy, superior subconjunctival mass was present. The cornea, anterior chamber, and iris of the left eye were normal, although a pink-white fundus reflex was present. Dilated fundus examination of the right eye revealed no abnormalities. The left eye had a large, elevated, pink-white subretinal tumor adjacent to the optic nerve and involving the nasal and superior-temporal quadrants (Fig 2). The subretinal tumor appeared very similar to an exophytic retinoblastoma. The central macula was not involved with tumor, although macular edema seemed present. The patient was admitted for further evaluation.

The admission general physical examination showed multiple subcutaneous masses over the scalp, several rubbery posterior cervical nodes, hepatomegaly, splenomegaly, and a possible right renal mass. Admission laboratory studies showed an elevated leukocyte blood cell count of 21,100 cells/mm\(^3\) with blast forms present; moderate thrombocytopenia was present. A computed tomography (CT) scan of the head and orbits, as well as an abdominal ultrasound, were ordered. The CT scan revealed a left orbital mass surrounding the optic nerve and extending superiorly over the globe. Intraocular tumor was present, but no calcification was present (Fig 3). No central nervous system tumor was noted. The abdominal ultrasound showed an enlarged right kidney, but no adrenal mass was visualized. Bone marrow aspiration and biopsy showed blast cells (Fig 4), and immunophenotyping showed early pre-B cell acute lymphoblastic leukemia.

With the diagnosis of acute leukemia established, chemotherapy with vincristine, daunorubicin, and prednisone was started. Additionally, a single dose of 180 rads of electron beam therapy was given to the left orbit. Rapid reduction of the proptosis occurred. Follow-up ophthalmic examination on the fourth hospital day showed significantly decreased proptosis with resolution of the intraocular tumor. Follow-up CT scan at the same time showed significant orbital tumor regression (Fig 5).

The patient had a fair induction with chemotherapy, and the leukemia was controlled for 2 months. He then developed a relapse, which showed little response to several
additional cycles of chemotherapy over the next 4 months. The patient was then seen to evaluate for signs of central nervous system leukemia. Ocular examination showed a large choroidal leukemic infiltrate in the right eye. The patient died 2 weeks later. The eyes were not available for histopathologic examination.

**DISCUSSION**

Our patient demonstrated leukokoria with proptosis due to an extensive intraocular and orbital tumor. Although the orbital tumor seemed to be a leukemic chloroma, the massive subretinal tumor was not at all typical of leukemia. The subretinal tumor looked similar to an exophytic retinoblastoma; the proximity to the optic disk made us consider orbital extension of retinoblastoma as a possibility, although the intraocular tumor seemed too small to be associated with such a large orbital tumor. Given the redness of the patient’s left upper eyelid, the recent reports of retinoblastoma presenting as orbital cellulitis also made us suspicious of retinoblastoma. The lack of tumor calcification on CT scan made retinoblastoma seem unlikely, although two cases of diffuse retinoblastoma without detectable calcification on CT scan have been reported. Other possibilities included orbital rhabdomyosarcoma and metastatic neuroblastoma, but the clinical findings were not consistent with these diagnoses. Establishing the correct diagnosis in this patient required a bone marrow aspiration-biopsy with immunophenotyping of the blast cells. If the bone marrow biopsy was nondiagnostic, a conjunctival and/or orbital biopsy would have been required.

The most likely diagnosis turned out to be the correct diagnosis, leukemia. The elevated leukocyte blood cell count, the blast forms on the peripheral blood smear, the rubbery suboccipital nodes, and the subcutaneous scalp masses all suggested leukemia, although the fundus picture was atypical, causing diagnostic confusion. The rapid response of the infiltrates to chemotherapy and low dose orbital irradiation provided confirmatory evidence. As the treatment of leukemia and retinoblastoma differ signifi-
The patient's globes were not available. The fundus appearance was highly suggestive of an intraocular tumor, and the initial CT scan seemed to show tumor within the globe without significant compression of the sclera. Given that the choroid is the most common site of ocular involvement in histopathologic studies, it seems plausible that a massive choroidal infiltrate could account for the observed signs. Even if the intraocular tumor was a compression effect, the important point is that not all cases of leukokoria with proptosis represent advanced retinoblastoma.

REFERENCES