A rare case of hypereosinophilic syndrome presenting with unilateral proptosis and torticollis

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Hypereosinophilic syndrome (HES) is a spectrum of myeloproliferative disorder, which is characterized by persistent and marked blood eosinophilia and damage to multiple organs due to eosinophilic infiltration. Idiopathic HES is identified after ruling out all other causes of eosinophilia. Poor prognosis is usually associated with cardiac involvement and malignant transformation of blood cells. We report a rare case of HES in an 8-year-old boy who presented with unilateral proptosis and torticollis. The patient responded well to corticosteroid therapy with reduction of proptosis and torticollis and normalization of serum eosinophil count.

Key words: Eosinophilia, hypereosinophilic syndrome, torticollis

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Hypereosinophilia has been defined as eosinophil count ≥1,500 eosinophils/μL. Hardy and Anderson in 1968 first described hypereosinophilic syndrome (HES) as sustained overproduction of eosinophils. The term idiopathic HES as defined by Chusid et al. was characterized by three criteria: (1) eosinophil count greater than 1,500 cells/μL persisting longer than 6 months, (2) single or multiple organ system dysfunction attributable to cytotoxic injury by eosinophils, and (3) without an identifiable etiology to explain the eosinophilia. The mortality in untreated patients can be as high as 75%. Inspite of significant amount progress in the understanding of the pathogenesis of HES, our knowledge is insufficient in formulating a new comprehensive definition of HES based on the etiology. Only a handful of reports can be retrieved from the literature that describe ocular involvement in HES. We present a unique case of HES, wherein an 8-year-old boy presented with unilateral proptosis with torticollis and eosinophilia. The child responded well to steroid therapy.

**Case Report**

An 8-year-old boy was referred to us with protrusion of his left eye (LE) for 1 month and neck stiffness for 7 days by the consulting pediatrician. Protrusion of LE was insidious in onset, slowly progressive, and was associated with mild pain. There was no history of diplopia, postural variation, and visual disturbance associated with protrusion of the eye. Neck stiffness was associated with mild pain. His medical and family history was unremarkable.

On examination [Fig. 1], child was of average built, well oriented to surroundings and afebrile. Visual acuity in both eyes was 6/6, N6. Head tilt to right was present. Child was orthophoric. An axial proptosis of LE was noted and was recorded to be 24 mm with the Hertel’s exophthalmometer. There was fullness of periorbital area with mild tenderness. On palpation of left orbit, orbital margins were intact, though insinuation with fingers was not possible. Proptosis was noncompressible and nonreducible. Bruits, thrills, and pulsations were absent. Slit lamp examination and fundus examination were within normal limits. Pupils were round, regular, and reacting to light. Intraocular pressure (Goldman’s applanation tonometry) was 19 mmHg in the right eye and 22 mmHg in the left eye. On systemic examination, vitals were stable. Mild pallor was present. Peripheral blood showed eosinophils were 62% with absolute eosinophil count to be 4,082 cells/μL [Fig. 2]. ESR was raised to 102 mm. There was no increase in the number of blasts or atypical cells in peripheral blood smear. All other laboratory work was within normal limits. Stool examination showed cysts of *Giardia lamblia*. No space occupying lesion in orbit was visible in USG B-scan of the orbit. MRI of orbits [Fig. 3a and b] showed thickening of orbital septum and lacrimal gland enlargement with fuzzy outline in left orbit. An MRI scan of cervical spine [Fig. 3c] showed loss of cervical lordosis. His chest x-ray [Fig. 3d] revealed bilateral paracardiac lower zone opacities. Ultrasonography of the abdomen showed mild hepatomegaly. The high eosinophil count could not be explained by the *Giardia* infection therefore, a diagnosis of HES was made and the same was conveyed to the consulting pediatrician. The child was started on corticosteroids in tapering doses (with a starting dose of 1 mg/kg), antiallergic, and antiprotzoal medications. He responded well to the therapy. After 2 weeks of treatment, there was marked reduction in proptosis and neck stiffness [Fig. 4]. At 6 months follow-up there was complete resolution of torticollis with reduction in proptosis and normal eosinophils counts. The patient is on 6 monthly follow-up with the ophthalmologist and the pediatrician and undergoes routine blood examination at each visit. This case is a rare one because, to our knowledge, unilateral proptosis with torticollis in a patient of HES has not been reported in the literature.

**Discussion**

Almost any organ or tissue may be affected in HES but mostly clinical presentation has been found to be due to skin, heart, lung, and CNS involvement. In the present case, lung, liver, cervical spine, and lacrimal gland along with periorbital tissue were involved. The clinical presentation of patients with HES is diverse and depends on the severity of the disease and pattern of end-organ involvement. Ocular manifestations in HES remains poorly described, thus highlighting its rarity. In one such report of a patient suffering from HES, transient visual obscuration preceded the systemic manifestations of the disease such as the renal infiltration.

There are only few reports of ocular involvement in HES. These reports mostly show retinocochoroidal vasculature involvement due to increased blood coagulability. Involvement of periorbital tissue with eosinophilic infiltration has not been reported so far.

In the present case to identify possible cause for eosinophilia elaborate history taking, examination, laboratory work-up, and investigations were done. Only possible cause for eosinophilia in this case may be *Giardiasis*. However, *Giardiasis* is rarely associated with eosinophilia and extremely rarely associated with such a high eosinophil count.

Corticosteroids constitute the 1st line of treatment. Oral prednisolone acetate in 1 mg/kg dosing in present case showed rapid improvement in proptosis and torticollis. Second line drugs include hydroxyureas, cyclosporin, and interferon-alpha. Newer modalities of therapy include the use of imatinib mesylate, monoclonal anti-IL5 antibody, and stem cell transplantation after nonmyeloablative conditioning.

In the present case, there was multisystem involvement in the form of orbital, musculoskeletal, pulmonary, and hepatic dysfunctions. We could not fulfill criteria of persistent elevation of eosinophil count for 6 months as it was not practically feasible to observe the patient for 6 months without proper management.

**Conclusion**

HES is a multisystem dysfunction secondary to eosinophilic infiltration. Rare presenting features of torticollis and proptosis and rewarding response to treatment in freeing the child of symptoms and a possible cure is unique in the present case.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other
clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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