# Case Report

# Extra-medullary Hematopoiesis Causing Bilateral Optic Atrophy in Beta Thalassemia/Hb E Disease

Nimitr Ittipunkul MD*,
Tada Martin MD*, Rutchada Siriwanasan MD*,
Kanokporn Olanratanachai MD**, Jack Rootman MD***

** Department of Ophthalmology, Faculty of Medicine, Chiang Mai University, Chiang Mai, Thailand*
*** Department of Radiology, Faculty of Medicine, Chiang Mai University, Chiang Mai, Thailand*
**** Department of Ophthalmology, Faculty of Medicine, University of British Columbia, Vancouver, Canada*

***Objective:*** *To report a rare presentation of bilateral optic nerve compression from extra-medullary hematopoiesis in beta thalassemia/Hb E disease.*
***Design:*** *Interventional case report.*
***Clinical presentation:*** *A 13-year-old Thai girl was reported with slowly progressive bilateral visual loss due to optic neuropathy as a result of compression from extra-medullary hematopoiesis. Computed tomography of orbit and brain revealed a homogeneous enhancing midline soft tissue mass originating from the ethmoid and sphenoid sinuses along with extensive involvement of the skull.*
***Intervention:*** *Blood transfusion once every month and low-dose radiotherapy to sphenoid and ethmoid bones with some visual improvement.*
***Conclusion:*** *Extra-medullary hematopoiesis in beta thalassemia can involve paranasal sinuses and cause compressive neuropathy. In patients with beta thalassemia that required multiple blood transfusions with progressive visual loss, ectopic marrow compression of optic nerve should be ruled out.*

***Keywords:*** *Optic atrophy, Blurred vision, Extramedullary hematopoiesis, Thalassemia*

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Extra-medullary hematopoiesis (EMH) is the production of hematopoietic elements in areas other than the bone marrow of the proximal long bones, spine, pelvis, and sternum. This condition is common in thalassemia, sickle cell anemia, and myeloproliferative disorders. EMH can also occur virtually in any organ, but most commonly in areas of fetal hemoglobin production including liver, spleen, lymph nodes. The paranasal sinuses are rare sites for the ectopic production[1,2]. The authors report a case of optic nerve compression due to EMH of the midline paranasal sinuses in a patient with beta-thalassemia/Hb E disease.

**Case Report**

The patient was a 13-year-old Thai girl with beta thalassemia/Hb E disease who developed symptomatic anemia at 5 months of age. Her father was a carrier of beta thalassemia, her mother had only beta thalassemia, and her sister had beta thalassemia/Hb E disease. At the age of 5 years, the patient underwent splenectomy and was noted to have extra-medullary hematopoiesis of omentum. She had a blood transfusion every 3 months and her hematocrit was between 12%-22% range but she never received chelation therapy.

At the presentation, she had slowly progressive visual loss in both eyes noted for two months. Prior to this the patient reported good vision. The examination showed she had short stature, rodent-like face with mark pallor, a grade 2 systolic murmur, and hepatomegaly.

On ocular examination, her visual acuity was finger count at one foot both eyes with best correct visual acuity of 2/60 right and 5/60 left. The fundus examination showed +2 disc pallor in both eyes. Color

*Correspondence to : Ittipunkul N, Department of Ophthalmology, Faculty of Medicine, Chiang Mai University, Chiang Mai 50200, Thailand.*

vision test using Ishihara plates was 0/12 both sides. She had tunnel vision with constriction of her visual field bilaterally (Fig. 1). Eye movements and the rest of her cranial nerves were intact. Hemogloblin was 7.5 g/dl at admission.

A computed tomographic(CT) scan of orbits and brain showed a homogeneous moderately enhancing soft tissue mass within the marrow cavity of both ethmoid and sphenoid sinuses causing expansion and rarefaction. A mass extended into the intracranial compartment with compression of the optic nerves and inferior frontal lobes (Fig. 2). Enhancing soft tissue masses in both maxillary sinuses were also observed as well as extensive enlargement of the skull.

The diagnosis of optic nerve compression caused by extra-medullary hematopoiesis was made and the patient received a course of external radiotherapy (1300 cGy in 6 fractions in one week) to the ethmoid and sphenoid bones and blood transfusions once a month.

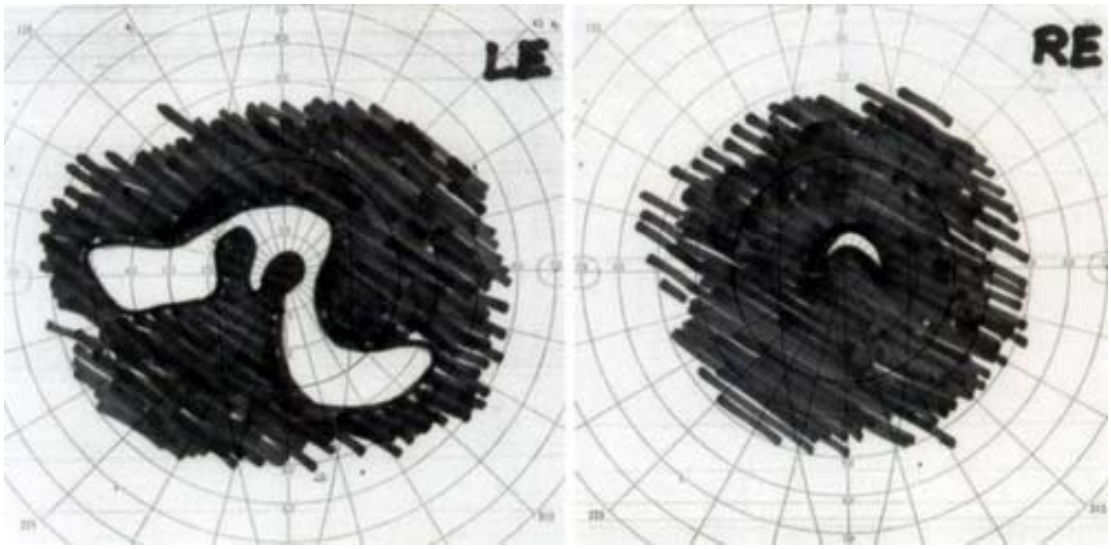


**Fig. 1** The visual field of both eyes shows severe constriction of the vision both sides

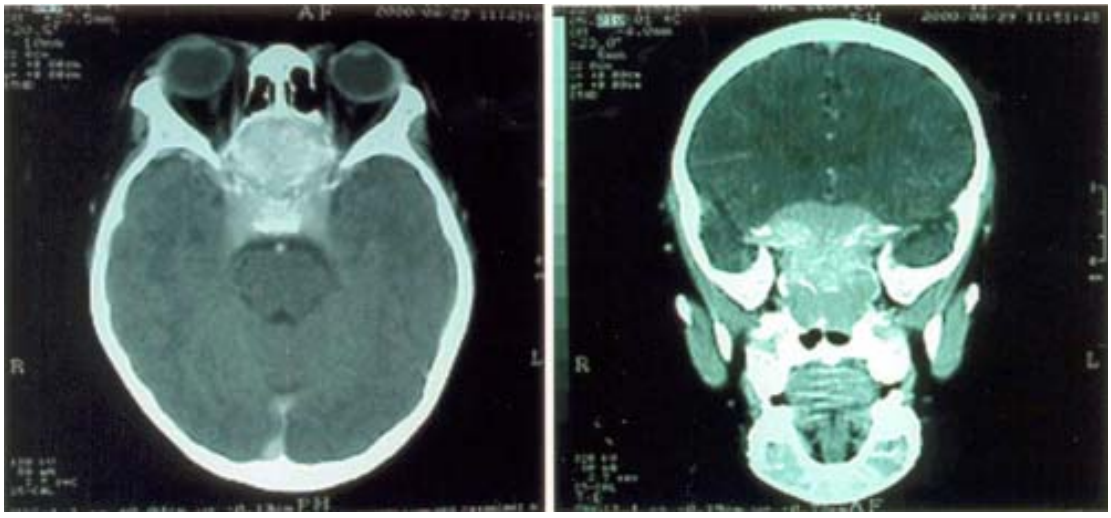


**Fig. 2** The axial and coronal CT scan of brain and orbit show a homogenous soft tissue mass within the ethmoid and sphenoid sinuses with compression of the optic nerves

The visual acuity improved at one week following the blood transfusion to 6/60 both eyes and after radiation was 6/36 right and 6/24 left.

**Discussion**

The thalassemia are a heterogenous group of heritable hypochromic anemias of varying degrees of severity classified as hemoglobinopathies. The underlying genetic defect results in decreasing or total suppression of hemoglobin polypeptide chain synthesis. They may be found in any ethnic population, but are most common in individuals from the Mediterranean area, Africa, the Middle East, the Indian subcontinent, and Southeast Asia

Homozygous beta thalassemia usually becomes symptomatic as a severe progressive hemolytic anemia during the sixth to twelfth month of life. Regular blood transfusions are necessary in these patients to prevent the profound weakness and cardiac decompensation. In untreated cases or in those receiving infrequent transfusions, hypertrophy of erythropoietic tissues occurs in medullary and extra-medullary locations. This patient had characteristic rodent-like face due to massive expansion of the marrow of face and skull.

Although the head and neck are unusual sites for EMH, there are a few reports involving maxillary, sphenoid, and ethmoid sinuses[1,2]. The presented patient had lesions in the intracranial compartment with bilateral compression of the optic nerves and inferior frontal lobes. The treatment modalities to relieve the pressure are varied including partial surgical decompression, low dose radiation, blood transfusion, and administration of hydroxyurea[2-6]. Because ectopic marrow is highly vascular tissue that can bleed easily, multiple transfusions to relieve anemia could lead to shrinkage of the ectopic marrow and extra-medullary marrow is highly sensitive to low-dose radiation[4,5]. The authors decided to avoid surgical intervention and gave low-dose radiotherapy combined with blood transfusion. The authors did not do a biopsy in this patient but diagnosed from the clinical finding and CT scan. The history of beta thalassemia/Hb E disease and splenectomy, the clinical hepatomegaly and the lesion in the ethmoid, sphenoid, and maxillary sinuses support the diagnosis of the extra-medullary hematopoiesis in the paranasal sinuses. The authors found that the patient's visual acuity had improved after blood transfusion and low-dose radiation.

Although the presentation as in this patient is rare, routine visual acuity check up in thalassemia patient may help for early detection of this abnormal finding.

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## ประสาทตาฝ่อจากภาวะ extramedullary hematopoiesis ในผู้ป่วยธาลัสซีเมีย

**นิมิตร อิทธิพันธุ์กุล, ธาดา มาร์ติน, รัชดา ศิริวันสาณฑ์, กนกพร โอฬารรัตนชัย, แจ็ค รูทแมน**

*ผู้เขียนได้รายงานผู้ป่วยเด็กหญิงไทยอายุ 13 ปีที่เป็นโรคธาลัสซีเมียชนิดเบต้าและฮีโมโกลบินอี (beta thalassemia/Hb E) ที่มาด้วยอาการตาทั้งสองข้างมัวลงมากขึ้นเรื่อย ๆ เนื่องจากประสาทตาฝ่อเพราะถูกกดโดย extramedullary hematopoiesis ที่มีขนาดโตขึ้นที่บริเวณ ethmoid sinus และ sphenoid sinus ผู้ป่วยได้รับการรักษาด้วยการให้เลือดและการฉายรังสี สามารถทำให้อาการตามัวดีขึ้นได้บางส่วน*