Extramedullary Haematopoietic Tumor Producing Small Intestinal Intussusception in a Beta-Thalassemia/ Hemoglobin E Thai Boy : A Case Report

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Abstract

The authors report a case of beta-thalassemia/hemoglobin E disease with extramedullary haematopoietic tumor which developed at the small intestine and caused intussusception. A 7 year-old boy with homozygous beta-thalassemia/hemoglobin E presented with recurrent abdominal pain. The abdominal ultrasonography showed ileo-ileal intussusception with a solid mass as the leading point. Resection of the ileal segment was performed. Pathological examination revealed an extramedullary haematopoietic tumor forming an intraluminal polypoid mass, being the leading point of the intussusception. Extramedullary haematopoiesis in the intestinal tract is rare. To our knowledge, this is the first case of extramedullary haematopoietic tumor that produced intussusception of the small intestine in a beta-thalassemia/hemoglobin E patient.

Key word : Thalassemia, Extramedullary Haematopoietic Tumor, Intussusception, Small Intestine

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Thalassemia is a form of hereditary anemia due to abnormality in hemoglobin production. Beta-thalassemia/hemoglobin E is the commonest form found in Southeast Asia[1]. In addition to increased intramedullary haematopoiesis, extramedullary haematopoiesis (EMH) is a common manifestation of severe thalassemia. The common sites of EMH are the liver, spleen and lymph nodes[2,3]. It may occur as paravertebral, intracranial, subcranial and subdural masses which could lead to serious

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complications from pressure effect[4-9]. Occasionally, some thalassemic patients present with an extramedullary haematopoietic tumor or tumors in the intrathoracic[10-12], mediastinum[13], liver[14, 15] and adrenal gland[16,17]. EMH in the small intestine is very rare. Five previously reported cases of intestinal EMH were presented with intestinal obstruction or intestinal hemorrhage from infiltration of the EMH and none of them occured in thalassemic patients[18-22]. Herein, we report a 7 year-old boy who presented with recurrent abdominal pain from reducible ileal intussusception. The leading point was an extramedullary haematopoietic tumor. Only one case of alpha-thalassemia has been presented with colo-colonic intussusception [23]. To our knowledge, this is the first reported case of EMH producing ileo-ileal intussusception.

CASE REPORT

A 7 year-old boy with homozygous beta-thalassemia/hemoglobin E was admitted due to recurrent abdominal pain for one month. He had been splenectomized at the age of 5 years and occasionally required blood tranfusion. His hematocrit was 21-24 per cent. Physical examination revealed a febrile patient with pallor. His abdomen was mildly distended with increased bowel sounds. A round firm smooth-surface suprapubic mass, 5 cm in diameter was palpated. The liver was palpable 3 cm below the right costal margin. Plain film of the abdomen showed intraluminal bowel mass with signs of gut obstruction (Fig. 1). Ultrasonography of the abdomen showed hepatomegaly without a space taking lesion and normal echogenicity. A solid mass, 4.5 cm in diameter, was seen originating from the bowel wall with signs of intussusception (Fig. 2). Barium enema revealed a normal colon and ileocaecal valve. Ultrafast CT scan of the whole abdomen revealed a round mass originating from the wall of the small bowel, being the leading point of the intussusception and causing dilation of the proximal jejunal loops (Fig. 3A and 3B).

The abdominal pain did not improve after treatment, so exploratory laparotomy was done. Generalized small bowel adhesion was noted. There was an intraluminal mass, 4 cm in diameter at the ileum, 2 feet from the ileocaecal valve with adhesion band and small bowel walled-off. Lysis of adhesion and resection of the ileal mass were performed.

The specimen consisted of a segment of the small intestine, 16 cm long. In the middle por-

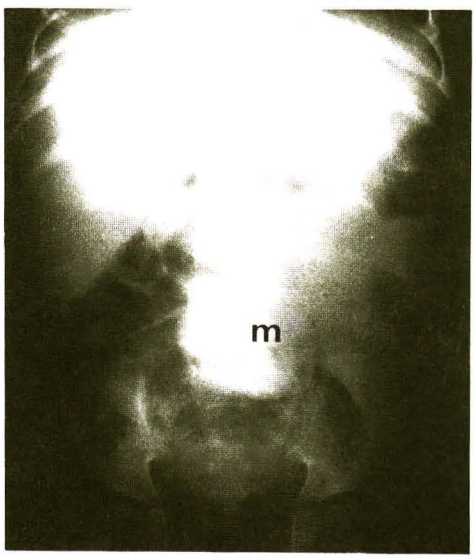

Fig. 1. Intraluminal bowel mass at mid abdomen causes mild degree gut obstruction. Supine film of abdomen shows crescent of bowel air partially surround the mass (m).

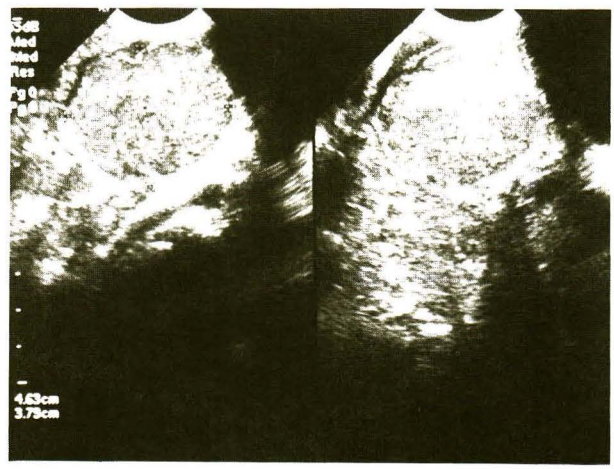

Fig. 2. Well-defined border soft tissue mass originating from bowel wall is the leading point of intussusception.

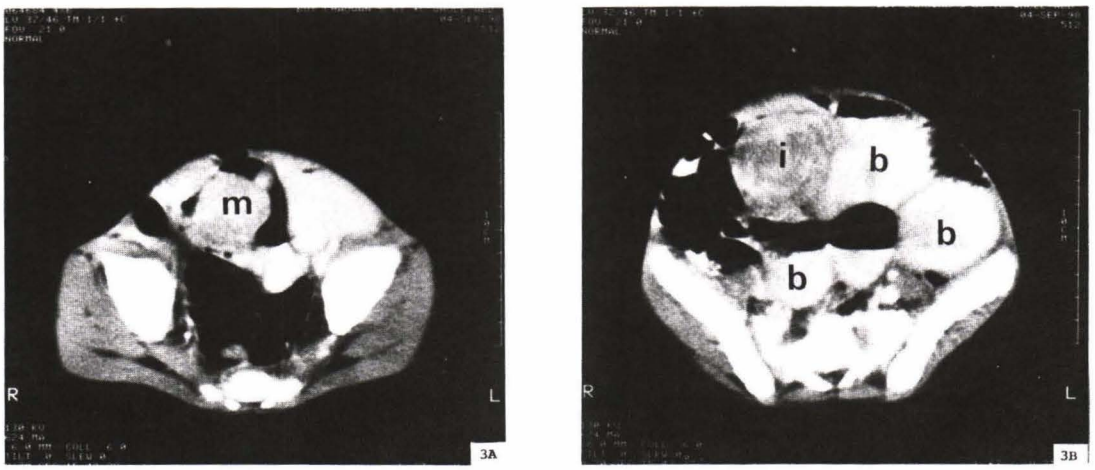

Fig. 3. (A, B) CT scans of abdomen shows the same findings.

A. CT scan at lower abdomen shows round soft tissue mass originating from wall of small bowel (m). It is the leading point of intussusception.

B. CT scan at upper level shows intussusception (i) with obstructive small bowel dilatation (b).

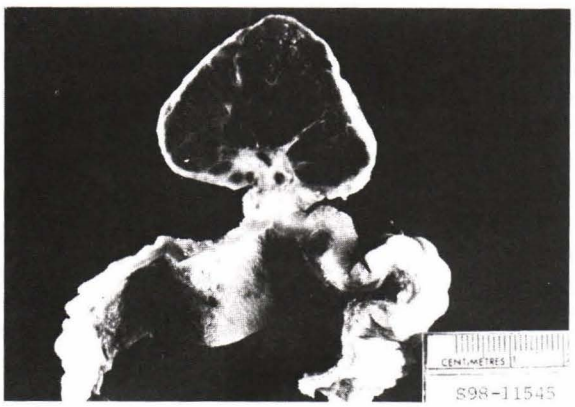

Fig. 4. The resected segment of ileum shows two lobulated polypoid masses with short stalk. They are the leading point of the intussusception. The cut surface of the mass is dark rusty red brown with fibrotic trabeculation. The covering mucosa is ulcerated and granulated.

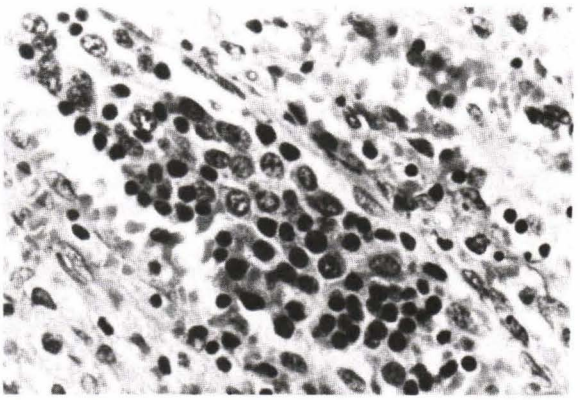

Fig. 5. (H&E x400) The higher magnification of the mass shows clusters of haematopoietic tissue, including the erythroid and myeloid series infiltrating in the fibrous stroma. Megakaryocyte is also demonstrated in this extramedullary haematopoietic tumor.

tion, there was a polypoid mass, 4.5x4x3 cm in size, with granular, lobulated, gray tan external surface. The cut surface showed a dark rusty red brown, firm submucosal mass with serosal extension. The mass showed fibrosis of the center near the base forming a short stalk. Another well circumscribed submucosal mass, 1.5x1x1 cm in size was seen adjacent to the first polypoid mass. It was only 1.5 cm from the first polypoid mass and had homogeneous dark red brown appearance on its cut surface (Fig. 4). Both masses showed extramedullary hematopoiesis with majority of erythroid

series. Myeloid series and megakaryocytes were also seen (Fig. 5). Fibrosis with hemosiderin deposition was noted. The superficial mucosal surface of the polypoid mass showed ulceration with granulation tissue formation and area of reepithelization near the base. The small mass showed intact mucosa. After the operation, he improved and was discharged without any complication.

DISCUSSION

Beta-thalassemia/hemoglobin E is a common hematologic disease in Thailand[1]. EMH frequently occurs in severe thalassemic patients and most commonly occurs in the liver, spleen and lymph node and less frequently in other organs such as kidneys, adrenal glands, heart, lungs and retroperitoneum[2,3]. Clinical manifestations secondary to EMH are variable and depend on the size and site of the hematopoietic tissue. An EMH forming tumor mass is uncommon but may cause serious results. Fucharoen et al reported two cases of intracranial EMH in beta-thalassemia/hemoglobin E patients[4,5]. One had epilepsy[4] and the other had brain atrophy from pressure effect[5]. Dhechakaisaya et al reported a case of EMH at the cranial dura mater and choroid plexus which caused convulsion in a thalassemia hemoglobin E disease[6]. Issaragrisil et al reported 12 cases of spinal cord compression in thalasssemia[7]. Among these serious neurologic complications, spinal cord compression from EMH seems to be most common[7-9]. Extramedullary haematopoietic tumor has occasionally been presented as tumors in intrathorax[10,11], posterior mediastinum[12,13], liver[14,15] and adrenal glands[16,17]. These presentations cause difficulty in diagnosis. EMH occuring in the gastrointestinal tract is very rare. There are only 3 cases of EMH occuring within the wall of the small intestine[18-20], one case at the caecum[21] and one at the rectum[22]. All cases occurred in patients with myelofibrosis. The presenting symptoms included hemorrhage[19,20], intestinal obstruction[18,19] and symptomatic constipation[22]. Our case was a thalassemic patient, presenting with signs of partial intestinal obstruction. Ultrasonography and whole abdomen ultrafast CT showed reducible ileal intussusception. The pathological examination of the resected ileum showed that the leading point of the intussusception was a polypoid extramedullary haematopoietic tumor.

Intussusception is the most common abdominal emergency in early childhood and the second most common cause of intestinal obstruction after pyloric stenosis[24]. Most of the cases are idiopathic in origin and only 2 per cent to 12 per cent of the cases from reported large series had a pathological lesion at the leading point identified[25,26]. This tends to occur more commonly in children older than 5 years of age[25]. Meckel's diverticulum is the most common cause in many series [24,26-31]. Our case seems to be the first case of extramedullary haematopoietic tumor that involved the small intestine of the beta-thalassemia/haemoglobin E patient and also the first case of intussusception with the leading point being an extramedullary haematopoietic tumor. There was only one 18 year-old Thai male with alpha-thalassemia having colo-colonic intussusception lead by an extramedullary haematopoietic mass of the colonic wall[23].

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การสร้างเม็ดเลือดนอกไขกระดูกทำให้เกิดลำไส้เล็กกลืนกันในเด็กชายไทยที่เป็นโรคเบต้า ธาลัสซีเมียฮีโมโกลบิน อี : รายงานผู้ป่วย 1 ราย

ศันสนีย์ วงศ์ไวศยวรรณ, พ.บ., รัตนพร พรกุล, พ.บ.**, สุเมธ ธีรรัตน์กุล, พ.บ.***, สามารถ ภคกษมา, พ.บ.****, สุภร ตรีพงษ์กรุณา, พ.บ.****, ประวัติ นิธิยานันท์, พ.บ.**

ได้รายงานผู้ป่วยโรคเบต้า ธาลัสซีเมีย ฮีโมโกลบิน อี 1 ราย ที่มีการสร้างเม็ดเลือดนอกไขกระดูกขึ้นที่ลำไส้เล็กส่วนปลายและทำให้เกิดลำไส้กลืนกัน ผู้ป่วยเด็กชายไทยอายุ 7 ปี เป็นโรคเบต้า ธาลัสซีเมีย ฮีโมโกลบิน อี มาโรงพยาบาลด้วยเรื่องปวดท้องเป็น ๆ หาย ๆ มาประมาณ 1 เดือน จากการตรวจช่องท้องด้วยวิธีอุลตร้าซาวนด์ พบว่ามีลำไส้ส่วนอิเลียมกลืนกันโดยมีตัวนำเป็นก้อนทูม ผู้ป่วยได้รับการผ่าตัดลำไส้เล็กส่วนอิเลียมออก ผลการตรวจทางพยาธิวิทยาพบว่าตัวนำของลำไส้กลืนกันเกิดจากการสร้างเม็ดเลือดนอกไขกระดูกจนมีลักษณะเป็นก้อนทูม การสร้างเม็ดเลือดนอกไขกระดูกที่ผนังลำไส้พบน้อยมาก เท่าที่มีรายงาน ผู้ป่วยรายนี้เป็นรายแรกที่มีการสร้างเม็ดเลือดนอกไขกระดูกจนเป็นสาเหตุทำให้เกิดลำไส้เล็กกลืนกัน

คำสำคัญ : ธาลัสซีเมีย, การสร้างเม็ดเลือดนอกไขกระดูก, ลำไส้กลืนกัน, ลำไส้เล็ก

ศันสนีย์ วงศ์ไวศยวรรณ และคณะ
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