· Case report ·

A case of hemophilia A with spontaneous fundal hemorrhage as its initial presentation

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Abstract

• Hemophilia is a rare disorder that occasionally has visual symptoms after diagnosis. The authors present a case in which visual symptoms led to the initial diagnosis, a 22-year-old man who represented with spontaneous fundal haemorrhage. There is no history of bleeding and trauma. The fundal haemorrhage was absorbed significantly without any treatment after 3 months follow-up. Hemophilia A was found to be the causative disorder in this previously healthy man. The authors present a rare case in which unilateral visual complaints led to the diagnosis of hemophilia A. Thus, though uncommon, hemophilia should be considered in the differential diagnosis of otherwise unexplained fundal haemorrhages.
• KEYWORDS: fundal haemorrhage; hemophilia A; retinal
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INTRODUCTION

Hemophilia A is a genetic bleeding disorder. Because of lack of clotting factor Ⅷ, the most common manifestation is bleeding in joints, gums, encephalon, and gastrointestinal tract, especially after trauma. The disorder occasionally has visual symptoms even after diagnosis. We present a case of hemophilia A in which visual symptoms led to the initial diagnosis.

CASE REPORT

A 22-year-old Chinese man visited a public hospital (Anhui Provincial Hospital) presented with a black shadow in visual field of left eye for 2 days and no obvious predisposing causes. He was the only child of consanguineous parents, and there is no bleeding tendency in his family. He had no history of trauma, haematological diseases, diabetes, vascular disease, or medication including anticoagulants. His best-corrected visual acuity of left eye was 0. 1. The anterior segment was normal, intraocular pressure was 15mmHg, but the optic disc was shaded by radial peripapillary haemorrhage except the temporal edge. Fundal examination also showed gross pieces of subretinal haemorrhage and scattered blutpunktes around macula lutea. These might be related with his poor best-corrected visual acuity. However, the abnormalities were limited to the left eye (Figure 1). On physical examination, there was no mucocutaneous haemorrhage, tenderness in sternum, splenohe-patomegalia, and other positive finding. Nevertheless, the laboratory findings revealed a prolonged activated partial thromboplastin time of 75 seconds (normal range, 30-50 seconds). Other biochemical (hepatic and renal function) and hematological evaluation (prothrombin time, international normalized ratio, thrombin time, and fibrin level) was normal.

It was thought he might have hemophilia. The clotting factor VIII and XI was checked, and the result showed a decreased factor VIII level of 2. 8% (normal range, 60%-150%) and a normal factor XI level of 87. 5% (normal range, 60%-150%). Blood Von Willebrand factor test was negative. Blood factor VIII-related antibody test was negative. A hematolo-gist was consulted that acquired hemophilia and Von Willebrand disease was excluded, and hemophilia A of mild type was diagnosed. The hematologist advised factor VIII infusion as needed perioperatively, besides that, no coagulation factors and drugs were needed for the moment. We concluded that the haemorrhage could absorb spontaneously. No therapy was performed, including medication and operation. After 3 months follow-up, the fundal haemorrhage was absorbed significantly, and visual acuity of left eye recovered to 0. 3.

DISCUSSION

Hemophilia A is a rare, genetic bleeding disorder. Because of lack of clotting factor VIII, the most common manifestation is bleeding in joints, gums, encephalon, and gastrointestinal tract, especially after trauma. Ocular complications (such as bleeding in eyelid, iris, choroids or retina) could happen with hemophilia, but hemophilia with spontaneous fundal haemorrhage as its first manifestation is extremely rare, no matter whether it is congenital or acquired.

In fact, most intraocular haemorrhage, though rare, happens after trauma. There are only a few reports concerning spontaneous fundal haemorrhage. Hon *et al*[1] reported an 80-year-old Chinese man of acquired hemophilia who presented with sudden haemorrhage in macula lutea. Wang *et al*[2] reported a 13-year-old boy with congenital hemophilia A with haemorrhage in optic disc after trauma. As to intraocular haemorrhage, though conservative treatment is enough in most patients, ocular surgery can be safely carried out with support of powerful haemostatic agent, such as recombinant activated

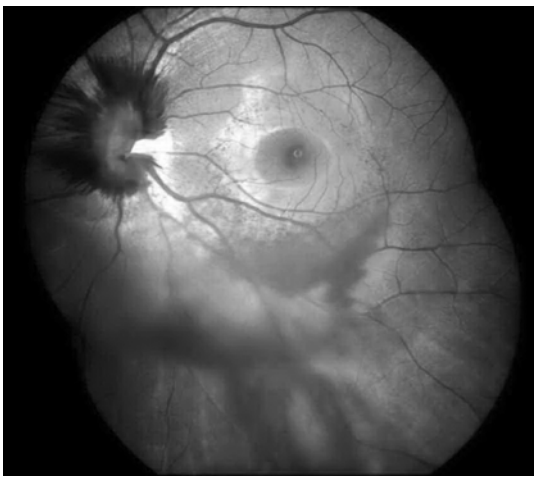

Figure 1 Fundus of the left eye of the patient with hemophilia A.

factor VII or factor VIII concentrates[1]. Nevertheless, there is not any precedent of surgery in fundal haemorrhage. In this case, since the bleeding was limited, and there was no vitreous haemorrhage or other complication, surgery was not considered.

As with all genetic disorders, a human can acquire hemophilia spontaneously through mutation, rather than inheriting it. Because no family history and other symptom of clotting disorder were found, fundal haemorrhage was the first presentation of this patient. It indicates that, in patients with spontaneous fundal haemorrhage, especially young patients without obvious predisposing causes, we must consider the possibility of hemophilia. Before diagnosis, if the bleeding got worse, it would be hard to control, just like the case with traumatic hyphema that Al-Fadhil *et al*[3] reported. Diagnosis in time and appropriate treatment can be helpful to avoid severe intraocular complications and improve the prognosis of these patients.

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以眼底出血为始发表现的甲型血友病 1 例

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摘要

血友病是一种少见的疾病,即使在诊断后也很少发生视力障碍。我们介绍 1 例因为视力障碍导致了血友病。男,22 岁,出现了自发性的眼底出血,没有外伤和出血史,未治疗,随访 3mo 后眼底出血明显吸收。这名男性青年在被诊断甲型血友病前完全健康。单眼视力障碍导致了甲型血友病。所以,在无法解释的眼底出血时,血友病应作为鉴别诊断之一来考虑。

关键词:眼底出血;甲型血友病;视网膜