

Case Report

Spontaneous Acute Epidural Haematoma in a Paediatric Patient with Congenital Afibrinogenaemia

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Abstract

Congenital afibrinogenaemia, a rare disorder of coagulation, is characterised by the congenital absence of fibrinogen. Uncontrolled umbilical cord stump bleeding is usually the first manifestation of the disease. Although various spontaneous haemorrhages may occur during lifetime, spontaneous epidural haematoma is rare. We report a case of congenital afibrinogenaemia in a paediatric patient who admitted to the emergency department with a complaint of headache. Computerised tomography scan of the head revealed a right parietal acute epidural haematoma. The patient underwent surgical evacuation subsequent to fibrinogen replacement in the emergency room. We decided to present our case since congenital afibrinogenaemia is rare in the emergency care. This report underlines congenital afibrinogenaemia as a potential cause of spontaneous acute epidural haematoma in paediatric patients which may require emergent intervention before the development of irreversible neurological conditions.

Key words

Congenital afibrinogenaemia; Paediatric emergency; Spontaneous epidural haematoma

Introduction

Fibrinogen (coagulation factor 1) is a large, fibrous plasma glycoprotein that plays a role in blood clot formation and platelet aggregation after its transformation into fibrin by thrombin.¹ Congenital afibrinogenaemia, a rare bleeding disorder showing an autosomal recessive pattern of

inheritance, is caused due to the congenital absence of fibrinogen. Uncontrolled umbilical stump bleeding in a newborn is the most common presentation and usually considered as diagnostic in congenital afibrinogenaemia. Although patients may present with spontaneous haemorrhages from variable regions of the body, spontaneous intracranial haemorrhages are rare. Moreover, spontaneous epidural haematomas beyond the neonatal period are even rarer.^{2,3} We report a case of a paediatric patient with spontaneous epidural haematoma who was presented to the emergency department and underwent successful surgical evacuation of the haematoma after the administration of intravenous (i.v.) fibrinogen concentrate in the emergency room. The rarity of this condition in emergency care prompted us to present this case.

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Case Report

An 8-year-old male patient with a history of congenital afibrinogenaemia presented to the emergency department with a complaint of headache, which had started several hours ago and worsened gradually. There was no history of

trauma. The patient had already been diagnosed with congenital afibrinogenaemia following massive umbilical stump bleeding in the neonatal period; however, this diagnosis was not further confirmed by genetic tests. Parental consanguinity was negative, and he had a healthy older sister. No other family members showed the signs of a similar disease. He had experienced frequent events of gingival bleeding and suffered from two episodes of patellar haemorrhage requiring conservative treatment 2 and 4 years ago. His vital signs and systemic physical examination were within normal limits. Glasgow coma scale revealed a score of 15, and there were no localising signs. He had isochoric pupils, with positive direct and indirect light reflexes. Prothrombin time and partial thromboplastin time levels were too long to be measured. Fibrinogen level could not be measured by functional assay. Computerised tomography scan of the head revealed a right parietal acute epidural haematoma that was nearly isodense to the brain parenchyma with a midline shift of 0.5 cm (Figure 1a, b). The patient underwent emergent craniotomy for acute, life-threatening epidural haematoma subsequent to i.v. administration of 100 mg/kg fibrinogen in the emergency room. Successful haematoma evacuation was confirmed by computerised tomography scan of the head (Figure 1c). The patient made excellent recovery, and he was discharged home on post-operative day 3 without any neurological deficits.

Discussion

Congenital afibrinogenaemia is a rare, autosomal recessive inherited bleeding disorder caused due to low or absent levels of fibrinogen in the patient's serum with an incidence of 1–2/1,000,000.¹ The clinical picture is variable exhibiting umbilical cord bleeding, haemorrhage into the muscle and joints, gingival haemorrhage, gastrointestinal and genitourinary bleeding, mucosal bleeding, splenic rupture and thrombotic events.^{2,4} Haemorrhages usually follow minor trauma or surgery, which was not the case in this patient. Spontaneous haemorrhages may occasionally occur. Intracerebral haemorrhage is a rare manifestation of congenital afibrinogenaemia, which can be lethal.⁵

Coagulation tests such as prothrombin time, partial thromboplastin time, thrombin time and bleeding time show prolonged duration. Fibrinogen is undetectable in patients' plasma (<10 mg/dL), which is diagnostic for the disease. Functional and immunological assays reveal absence or very low levels of fibrinogen.

Afibrinogenaemia is characterised by a tendency to bleed. However, patients may have long time intervals between bleeding periods. Our patient had experienced two episodes of patellar haemorrhage due to minor trauma 2 and 4 years ago since the time of diagnosis of congenital afibrinogenaemia immediately after birth. Although the majority of bleeding events occur subsequent to trauma or

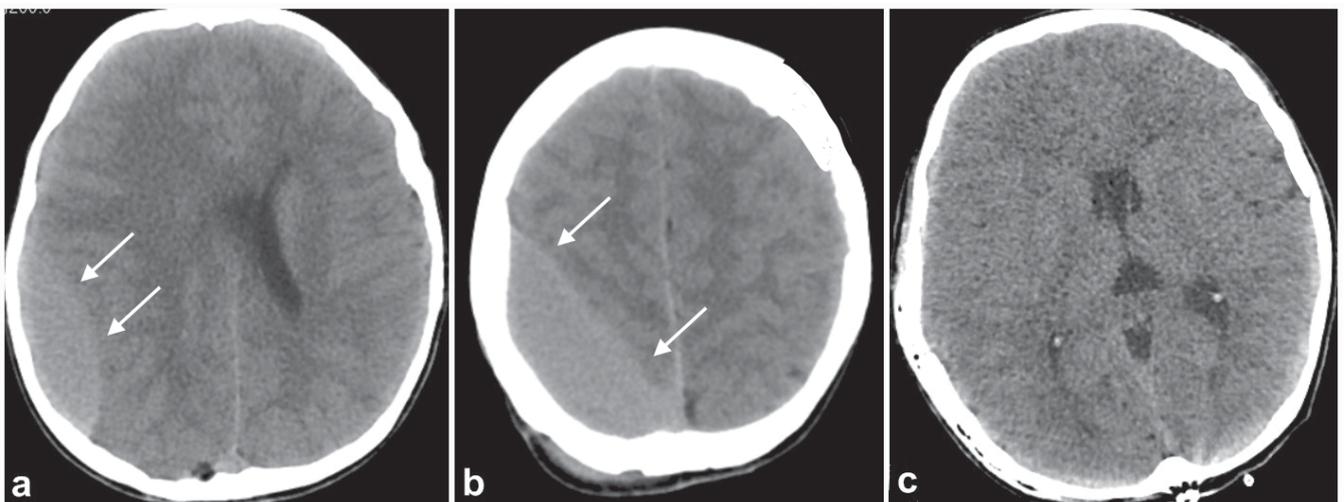


Figure 1 (a) and (b) Computerised tomography scan of the head revealing a right parietal acute epidural haematoma (white arrows). (c) Computerised tomography scan of the head of the same patient demonstrating complete evacuation of the haematoma following surgery.

surgery, spontaneous haemorrhages may be encountered occasionally.

In general, epidural haematomas are associated with head trauma. A spontaneous epidural haematoma is an uncommon occurrence that is primarily observed in patients with chronic renal failure or during open heart surgery, middle meningeal artery aneurysm, systemic lupus erythematosus,⁶ sickle cell disease, haemorrhagic tumours, dural vascular malformations, blood coagulation disorders or infections of the adjacent regions such as otitis and paranasal sinusitis.⁷ Epidural haematomas, occurring due to any cause, may become life-threatening with rapid deterioration of the neurological status of the patient, leading to high morbidity and mortality rates if not recognised in time.

Spontaneous epidural haematoma is a rare manifestation of congenital afibrinogenemia in the paediatric population, as well. Hence, knowledge regarding spontaneous epidural haematomas in these patients does not extend beyond case reports according to the published literature.^{2,3} To the best of our knowledge, our case is the third paediatric patient with congenital afibrinogenemia who presented with spontaneous epidural haematoma and underwent successful surgical evacuation.

Intracerebral haemorrhages in patients with coagulation disorders may be undetectable or isodense due to the clotting defect.⁸ Therefore, computerised tomography scans might overlook even frank haemorrhages in the initial phase, thus requiring further evaluation and surveillance of the patient. Absence of hyperdensity and probability of isodense haemorrhagic lesions on computerised tomography scans occurring due to the clotting disorder, requiring careful and thorough examination of the images, must also be considered in patients with bleeding disorders.

Fibrinogen replacement is the treatment of choice in patients with afibrinogenemia with intracranial bleeding prior to surgery to avoid perioperative haemorrhages precluding haemostasis and safe surgery. Plasma fibrinogen levels more than 0.5-1.0 g/L are also recommended to be established for 4-14 days following surgery.⁹ Cryoprecipitate and fresh frozen plasma are the agents that must be used in emergent cases when fibrinogen is not available.¹⁰ Our patient was treated with i.v. fibrinogen prior to surgery as it was available. Prophylactic treatment is still of debate due to the potential thromboembolic complications and side effects of blood products.

Conclusion

Emergency care providers should be aware of the spontaneous occurrence of intracranial haemorrhages in patients with congenital afibrinogenemia. The clinical diagnosis largely depends on suspicion and appropriate neuroimaging due to the probability of unexpected and life-threatening intracranial haematomas. Moreover, acute haematomas may be undetectable or isodense to the brain parenchyma, thus rendering the diagnosis more difficult in afibrinogenemic patients. Fibrinogen, fresh frozen plasma or cryoprecipitate may be used in emergent medical care, which may also provide safe surgery. Consultation with a paediatric haematologist may be a reasonable option in planning the dose and timing of the treatment. These approaches can help in achieving an early diagnosis and successful treatment in paediatric patients with spontaneous haematomas before the development of irreversible neurological conditions.

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