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CASE REPORT

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δ-Storage pool disease: a pitfall in the forensic investigation of sudden anal blood loss in children: a case report

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Abstract We present the case of a 3.5-year-old boy with sudden anal blood loss at school. Sexual abuse was suspected, and, apart from anal fissures seen on sigmoidoscopy, no other clinical signs of any sort of disorder were present. As no medical explanation for the blood loss could be given, penetrating anal trauma was suggested. During follow-up consultations, there were complaints of occasional blood loss. Platelet aggregation tests and electron microscopy finally helped diagnose a δ-storage pool disease which is a rare haemostatic disorder involving the dense granules of the platelets. Although exclusion of well-known blood diseases through routine laboratory testing is a common practice in children with sudden blood loss, this case illustrates the value of more specialised investigation both from a diagnostic and forensic point of view.

Keywords Children · Sexual abuse · δ-Storage pool disease · Platelets · Electron microscopy

Introduction

Sudden anal blood loss in children is always a diagnostic challenge. Not only is anal blood loss a very frightening sensation for a child that demands immediate care but it should also direct towards a more forensic-oriented approach to preserve and document possible signs of abuse, especially when a consistent explanation for the event is missing and when other clinical signs of possible abuse are present.

It is general knowledge that in the differential diagnosis of a child with multiple bruises all over its body, haemostatic disorders should be included as well as child abuse. The Ehler–Danlos syndrome is one of the well-documented connective tissue diseases associated with a high incidence of haematoma. Clotting diseases and platelet disorders should also be considered as well as diseases associated with pigment malformation. A thorough clinical investigation and routine blood tests usually help to diagnose these diseases.

To our knowledge, we present the case of first δ-storage pool disease that was diagnosed in a young child with anal blood loss as presenting symptom, demanding forensic intervention.

Case report

A 3.5-year-old boy was brought to the emergency department by his kindergarten teacher. A couple of hours before admission, he had returned from a visit to the bathroom smothered in blood, his underwear soaked in blood. According to the school personnel, the toilet walls were also covered with bloodstains.

In the emergency department, both doctors and teacher were alarmed about a possible sexual abuse situation and decided to alert the police.

Anamnesis of both teachers and parents brought conflicting information. The last couple of days, the boy had been very unwilling to go to school. He had complained of anal itching. The boy had a very shy nature and hardly said a word; he only told a story of being beaten with a spoon, but it was unclear by whom, where and when. As the boy was always brought to school by his father who also assisted him to the toilet before taking off, the father was the first suspect.

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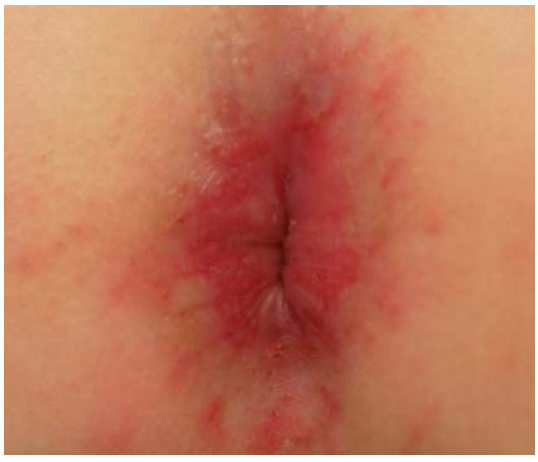

Fig. 1 Anal clinical investigation. Anal view in supine position: note the superficial fissures, the soiling, the perianal erythema and the skin erosions

Clinical investigations of the heart, lungs and abdomen were normal, and no skin lesions could be demonstrated. The boy was very distressed during the investigation. Anal investigation in both prone and supine position revealed a slightly distended anus, circumferentially irritated, with several superficial mucosal erosions (Fig. 1). Very little incrusted blood could be seen. No worms were found. The boy had been washed before coming to the hospital, and in his underwear, only small blood streaks were seen.

At sigmoidoscopy, the boy was extremely agitated and frightened although he was sedated with 3 mg midazolam and 12.5 mg pethidine. Five small anal fissures were found. The rectal mucosa was hypervascular over a distance of 15 cm; no internal cause for the blood loss could be seen.

Routine laboratory blood analysis on admission revealed a normal haemoglobin count (11.5 g/dl), a normal platelet count (280×10^9/l) and liver and renal function tests within normal range. Coagulation studies revealed a normal prothrombin time and prolonged activated partial thromboplastin time (aPPT): 75.5 s (nl 24–38 s). Additionally, testing for lupus anticoagulant was positive, which explained the prolonged aPPT but was no explanation for blood loss in the presence of a normal platelet count. No other specialised laboratory tests were performed. Faecal culture grew *Escherichia coli*. No parasites could be demonstrated.

Anamnesis in depth of both parents revealed that the boy occasionally had blood stains in his underwear. His mother reported having frequent nasal bleedings and his uncle was said to often have gum bleeding. There was no history of constipation, and also during his hospitalisation, this was no problem, according to the medical files.

During hospitalisation, two more episodes of anal blood loss were reported. Twice, there was a little bit of blood with his stool. No gastro-intestinal pathology could be demonstrated; an additional Meckel scan was negative. The boy left the hospital 2 weeks later for a child protection centre where he could be visited by his parents.

At interval consultations, the boy was in good physical condition, occasional discrete blood staining of his underwear being the only thing worth mentioning. At a second sigmoidoscopy, retraction of the scope created a rectal wall haematoma. Rectal mucosa was still normal. In the protection centre, the boy seemed to suffer from constipation occasionally; when given lactulose as laxative, his stool was softer and no further bleeding events occurred.

Although aggregation tests were normal, electron microscopy eventually revealed the diagnosis of δ-storage pool disease (Fig. 2).

The conclusion of the forensic investigation was that this boy suffered from a δ-storage pool disease, a platelet disorder. It was stated that this disease could explain ample anal blood loss after minor trauma (e.g. scratching, constipation, etc.), but that the peculiar behaviour of the boy and the anal fissures remained unexplained and could still be the result of penetrating anal trauma, although the presence of the above-mentioned pathology made the situation less suspect. As police investigation did not come up with other evidence, charges against the family were dropped and the boy was allowed to return home after 3 months.

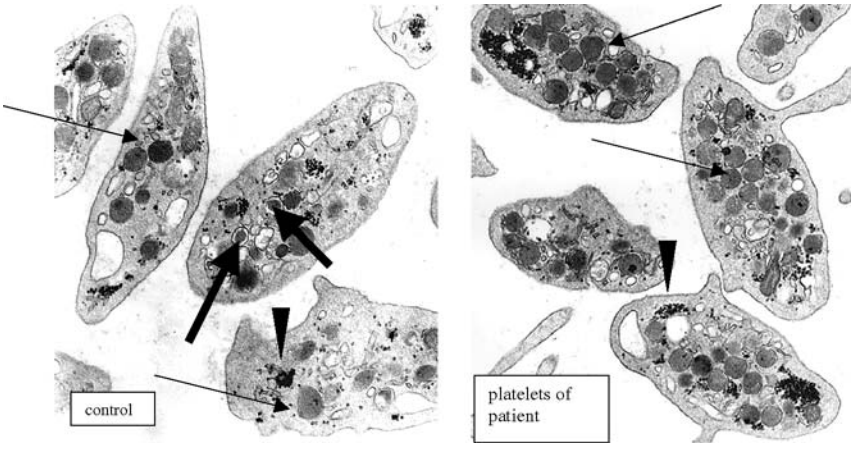

Fig. 2 Electron microscopic view of platelet with and without dense granules. Transmission electron microscopy of normal platelets (*left*) containing granules, α granules and lysosomes (*arrow*), as well as dense core granules (*thick arrow*) and glycogen particles (*arrowhead*). Transmission electron microscopy of the patient's platelets (*right*) containing α granules, lysosomes and glycogen particles. Dense core granules are absent. Original magnification ×20,000

Discussion

More and more medical professionals are aware of the importance of considering the diagnosis of sexual abuse within the context of the primary care practice [1]. Taking sexual abuse into the diagnostic process is an important step in correlating a pattern of behavioural and medical indicators with the possibility of sexual abuse [2]. Behavioural indicators include a history of a newly manifested clinging behaviour and irritability, thumb sucking and loss of bowel control, sleepwalking or bed-wetting, anorexia, loss of concentration and sexualised behaviour inappropriate for developmental age [3, 4]. Medical indicators are bruising, scratching, bites, sexually transmitted diseases, bloodstained underwear, bruising of the anogenital region, bruises without consistent history, enuresis, constipation and anal fissures. Genital examination should focus on the external genital region of both girls and boys [3].

Anal sexual abuse is mostly seen in very young children and in boys [5]. Almost always, some sort of violence has been used, although not always with visible traumatic lesions. Anal lesions are almost always non-specific and can only be seen shortly after the assault as the mucous lining has a rapid healing capacity [1, 3, 5, 6]. Anal examination can be done in supine, prone and left-lateral positions. The anal and perianal soft tissues and the sphincter tone have to be examined. Even in the absence of clinical signs, tests to detect spermatozoa should be systematically performed in any case of suspected assault, best within the first 24 h after the assault [7].

The differential diagnosis of anal blood loss in children should, apart from anal lesions, include food allergy, necrotizing enterocolitis, infectious enterocolitis, Hirschsprung's disease, Meckel's diverticulum, duplication of the bowel, intussusception, volvulus, polyps, lymphonodular hyperplasia, inflammatory bowel disease, angiodysplasia, haemorrhoids, Henoch–Schönlein purpura, haemolytic–uremic syndrome and bleeding diathesis such as δ-storage pool disease [8].

Platelet storage pool deficiencies comprise a range of disorders encompassing variable degrees of reduction in the numbers of both number and contents of dense granules (δ granules), α granules or both [9]. Dense granules were originally identified as storage organelles in platelets of mammalian species, containing in humans, serotonin, calcium, adenosine diphosphate (ADP), adenosine triphosphate and pyrophosphate [10–13]. When platelets are activated, these granules release the content through the canalicular system [14]. Release of ADP is responsible for the so-called second wave in the biphasic platelet aggregation curve. The dense granules play an important role in the propagation of the primary platelet response [14, 15].

Patients with decreased amounts of these substances stored in the platelet-dense granules have one of the most common forms of platelet function disorders: δ-storage pool deficiency (δ-SPD). Several types of δ-SPD have already been identified, associated with both acquired clinical conditions and congenital syndromes, e.g. Chediak–Higashi syndrome [10, 12, 13, 16].

δ-SPD was also diagnosed as a solitary finding in patients with a mild-bleeding diathesis presenting clinically with either easy bruising, epistaxis, menorraghia and/or postoperative blood loss. This entity is called δ-storage pool disease and is transmitted as an autosomal dominant trait [16, 17]. A variable degree of granule deficiency results in a wide range of clinical and laboratory manifestations.

The diagnosis of δ-storage pool disease is mostly based on the clinical presentation: sudden unexpected blood loss in various situations such as child birth or surgery. There seems to be a slight male predominance. The youngest patient in the literature diagnosed with this disease was 6 years old [4]. The late age at diagnosis is due to the usually harmless symptoms of this disease. Platelet count and platelet morphology are usually normal, although mild thrombocytopenia and increased platelet size can be seen as well as a prolonged bleeding time [9, 16, 18]. In platelet aggregation studies, commonly the second wave, stimulated by ADP, is missing in δ-storage pool disease, but cases with normal aggregation studies have also been reported [17, 18]. Complementary, for example, flow cytometry of mepacrine-treated platelets is also possible [17, 19]. However, as dense granules are inherently electron opaque probably due to their high-calcium content, they can readily be examined by electron microscopy [13]. This allows not only quantification of the number of dense granules but also evaluation of their morphology [16]. Four types of platelets have been identified in relation to their dense granule content, but the clinical relevance of the different types is not yet clear [11]. Total absence of dense granules has until now never been seen. The pathogenesis of δ-storage pool disease is currently not completely understood, and an association with a lacking membranous protein, granulophysin, has been suggested [12, 16].

No daily treatment is necessary. When extensive blood loss can be foreseen, e.g. in operations, treatment is available, e.g. inhibitors of fibrinolysis or platelet transfusions. Patients are advised to carry a tag indicating the disease, its symptoms and necessary treatment in case of emergencies.

Conclusion

δ-Storage pool disease is a platelet function disorder that only occasionally leads to sudden blood loss or bruising. It can only be diagnosed through platelet aggregation tests, platelet secretion tests and electron microscopy, as routine laboratory tests are usually within the normal range. The presentation of the disease in our case was for several reasons unusual. We were initially confronted with excessive anal blood loss in a young child, which raised the suspicion of sexual abuse and led to a forensic intervention.

However, we wish to emphasize that in the diagnosis of abuse, an overall consideration of the situation remains the priority: the diagnosis of this disease may explain certain features, but there can still be an abuse situation in the background as a haemostatic disorder is known to aggra-

vate the symptoms of sexual abuse and child abuse in general, including Munchausen syndrome by proxy [20].

We believe it to be worthwhile to exclude this disease and other haemostatic disorders in possible abuse situations. In case of minor haematological abnormalities and unexplained symptoms, it might be useful to perform electron microscopy. In a therapeutic setting, it offers an explanation for the blood loss, and on the forensic front, a probability range of different explanations for the symptoms can be established on which the police investigation can be based.

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