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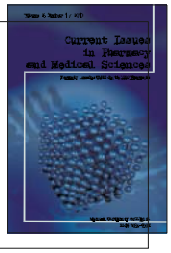

The circumstances of diagnosis of occult haemophilia B in a patient with severe maxillofacial injuries

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ABSTRACT

The most common haemostasis disorders are haemophilia A, B and von Willebrand disease. Symptoms of congenital bleeding diathesis can be observed in early childhood, and are easy to recognize. Medical literature describes cases of late diagnosis of haemostasis disorders. These cases are usually detected accidentally, and they can present a serious diagnostic and therapeutic problem. In this article, the authors describe a patient with massive maxillofacial trauma in whom a haemostasis disorder was detected and diagnosed as haemophilia B. Detection of this disorder was of significant importance for the treatment of the primary disease, i.e. maxillofacial injuries.

Keywords: fractures, maxillofacial injury, inherited bleeding discorders, haemophilia B

INTRODUCTION

The purpose of this article is to present the issue of late detection of haemostasis disorders and associated difficulties with diagnosis and therapy. Haemophilia is the most common congenital coagulopathy. It was first described by the Philadelphia doctor J. C. Otto in 1803. In Poland, the first reports regarding haemophilia date from the 1850s, whereas the 1950s brought the greatest progress in studies on its etiopathogenesis and new methods of treatment. Today, haemophilia is a relatively well-studied coagulopathy, and patients suffering from this disease receive medical care and specialist treatment from their youngest years [4, 6, 7].

The typical haemophilia diagnosis is made on the basis of a propensity for bleeding in joints and muscles since early childhood. The patient's medical history, together with measurements of coagulation factor levels, enables physicians to accurately determine the type and form of the coagulopathy. Thus, early detection and diagnosis of haemophilia in young children makes it possible to initiate factor replacement therapy and prophylactic treatment, as well as – in recent years – treatment at home, which has significantly improved these patients' quality of life. In Poland, as in other European countries, the treatment of patients suffering from haemophilia and other coagulopathies is overseen by state institutions. On the state level, treatment protocols and standards are controlled by the Institute of Haematology and Transfusion Medicine, while Regional Centers of Blood Donation and Treatment provide specialist care on the provincial and communal level [2, 5].

Medical literature shows that patients with mild and subclinical, often occult forms of haemostasis disorders can present a significant problem, and this is corroborated by everyday clinical practice. In such situations, diagnosing the coagulopathy is difficult and occurs accidentally, usually in adulthood. In these cases, bleeding episodes and symptoms of the haemorrhagic diathesis are atypical, usually accompanying small injuries and various, frequently minor surgical interventions, often dental surgery procedures [1, 3].

AIM

The purpose of this article is to present the issue of late detection of haemostasis disorders and associated difficulties with diagnosis and therapy.

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CASE RAPORT

In the Clinical Ward of Maxillofacial Surgery of the Provincial Specialist Hospital in Rzeszów, a patient was recently hospitalized for treatment of maxillofacial trauma incurred during a workplace accident. This patient was additionally diagnosed with haemophilia B.

A man aged 35 years was admitted to the Clinical Ward of Maxillofacial Surgery because of maxillofacial trauma sustained while operating a circular saw. He did not lose consciousness after the injury. He remembered the circumstances of the accident. The patient's general condition upon admission was good. Pronounced facial assymetry was noted, caused by an extensive haematoma of the right cheek and orbit. The upper and lower right eyelids were also involved, leading to eye closure. Heavy bleeding from the right nasal canal, the oral cavity and facial wounds proved difficult to control. The patient reported severe pain and feelings of pressure in the right half of his face, as well as increasing difficulty with jaw opening. He was diagnosed with soft tissue and skin injuries due to blunt force trauma and suspected comminuted fractures of the facial region of the skull. To make a definite diagnosis and determine the extent of bone damage, conventional radiological examinations and a CT scan of the head were performed. Medical imaging (radiological examinations) confirmed the presence of a zygomatic-mandibular-orbital fracture on the right side, with displacement of the bone fragments. The basic, routine laboratory blood tests were performed and, apart from leukocytosis and an elevated erythrocyte sedimentation rate and CRP level, the results were normal. Because of massive epistaxis and a trauma wound involving the patient's nose and right cheek, he was qualified for immediate treatment of facial soft tissue injuries under general anaesthesia. The facial wounds were closed surgically and the epistaxis was treated using a Bellocq tamponade.

During the postoperative period, despite meticulous surgical closure of the wounds and no evident pathological changes in subsequent laboratory blood tests, „seeping" bleeding from the postoperative wounds continued. This situation indicated a need to identify the cause of the bleeding and the presence of a bleeding diathesis was suspected. Data gathered via medical interview, together with a careful family history, provided no significant information which would help determine the cause of the bleeding. In the subsequent days of the patient's hospital stay, in addition to facial bruising, ecchymoses appeared on his neck and chest. The patient also reported blood „trickling" down the back of his throat. Because of a worsening general condition due to prolonged bleeding, as well as the fact that the cause of this bleeding remained unidentified, the planned surgical procedure to reposition and stabilize the bone fragments of the zygomatic-mandibular-orbital fracture was postponed.

Because of a suspected systemic cause of the bleeding and the suspicion of a bleeding diathesis, an extended panel of blood coagulation tests were performed, i.e. bleeding time, plasma recalcification time, activated whole blood clotting time, activated partial thromboplastin time, prothrombin time, fibrinogen concentration. The results showed a normal bleeding time (5min, 23 s) and a prolonged whole blood clotting time (21 min, 03 s), whereas the activated partial prothrombin time was 48,7 s. For this reason, the levels of coagulation factor VIII and IX were measured. The assays showed a decreased concentration of factor IX – only 11% of the normal level. After an analysis of the disease course and a haematological consultation, a diagnosis of occult, subclinical haemophilia B was made.

Since the patient required surgical treatment of his facial fractures, preparatory procedures were initiated in accordance with universally recognized standards of patient preparation for elective surgery. The plan was to elevate the concentration of the deficient factor IX to 80% of the normal level. Before the operation, the patient received 3400 units of factor IX concentrate. He tolerated the operation well, and no severe postoperative bleeding was observed. In the postoperative period, the patient received 1500 units of factor IX concentrate intravenously every 24 hours until the wound was healed and sutures were removed. Over the entire treatment period, a total of 13900 units of lyophilized factor IX were infused. The patient also received antifibrynolytic treatment in the form of tranexamic acid. The daily dosage was 200-350 mg per kilogram of body weight. The antifibrynolytic treatment was continued until the wound was healed. No heavy bleeding was noted during the surgery, which was performed in an atraumatic fashion. To inhibit intraoperative local bleeding, the fracture was covered with the fibrin-collagen hemostatic agent Tacho-Comb. Surgical repair of the zygomatic-mandibular-orbital fracture was achieved by repositioning the bone fragments in the first step of the procedure, and subsequently stabilizing them with miniature plates. No abnormal bleeding was observed during the postoperative period or during healing. The patient was discharged 10 days after the operation in a good general and local condition. In addition to a diagnosis of maxillofacial fractures, he was informed about the diagnosis of mild haemophilia B. A visit and consultation in the Regional Center of Blood Donation and Treatment were recommended in order to confirm this diagnosis and determine the type and form of the coagulopathy. During outpatient control visits, no bleeding incidents or complications in wound healing were noted. During ambulatory treatment, the patient did not confirm having fulfilled the recommendation of undergoing further diagnosis of his bleeding diathesis.

CONCLUSION

The above case of a late diagnosis of haemophilia B in a 35-year-old man with no previous clinical symptoms, in whom the disease only became apparent as a result of severe maxillofacial trauma, is in line with existing reports that haemostasis disorders, especially mild or asymptomatic forms, may be diagnosed as late as adulthood, and confirms the need for comprehensive diagnostics to determine the cause of prolonged bleeding. The diagnosis of a haemostasis disorder should always be confirmed by the Institute of Haematology and Transfusion Medicine in Warsaw in order to determine the type of the coagulopathy and the severity of coagulation factor deficiency [2, 3, 5, 7].

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