Unusual association of diseases/symptoms

Idiopathic facial swelling secondary to sickle cell anaemia

Swapnil Moghe,¹ Ajay Pillai,¹ Kanishka Navin Guru,² Preeti P Nair³¹Department of Oral & Maxillofacial Surgery, People's Dental Academy, Bhopal, Madhya Pradesh, India²Department of Oral Surgery, People's University, Bhopal, Madhya Pradesh, India³Department of Oral Medicine & Radiology, People's College of Dental Sciences and Research Centre, Bhopal, Madhya Pradesh, India

Correspondence to Professor Preeti P Nair, shajihoss@gmail.com

Summary

Sickle cell disease is a common inherited autosomal disease that is characterised by abnormally shaped (sickle-shaped) red blood cells (RBCs). It can involve virtually any organ system. The clinical manifestations of sickle cell disease vary and are classified as vaso-occlusion, chronic anaemia and infection. The imaging appearances of central nervous system and musculoskeletal involvement by sickle cell disease have been well documented; however, involvement of the head and neck region is often unreported, although it is not uncommon. In the head and neck, sickle cell disease can involve the inner ears, orbits, paranasal sinuses, bones, lymph nodes and vessels. This paper describes a case of idiopathic facial swelling associated with sickle cell disease in a young patient.

BACKGROUND

Sickle cell disease is a common inherited autosomal blood disorder that is characterised by the presence of sickle-shaped red blood cells. The clinical manifestations of sickle cell disease vary, but they may be attributed to three mechanisms: vaso-occlusion, chronic haemolytic anaemia and infection. It can involve virtually any organ system. The features of this condition affecting the head-and-neck region remains poorly reported. In the head and neck, sickle cell disease can involve the inner ears, orbits, paranasal sinuses, bones, lymph nodes and vessels.

CASE PRESENTATION

A 20-year-old male patient reported with a complaint of swelling in the left infraorbital region (figure 1). Thorough clinical and radiographic examination was inconclusive with regard to the cause of swelling. The patient came on the following day with complaint of swelling over right cheek region (figure 2). Investigation revealed that the patient suffered from sickle cell disease. On the basis of this diagnosis, the swelling could be attributed to shifting of emboli leading to swelling in a particular area of the body.

INVESTIGATIONS

- ▶ Intraoral periapical radiograph in relation to 23, 24 and 25 which revealed no pathology.
- ▶ Orthopantomogram revealed no aetiology of dental origin (figure 3).
- ▶ Complete blood count, Hb as 6 gm%.

DIFFERENTIAL DIAGNOSIS

Anaemic disorders associated with orofacial signs and symptoms include iron deficiency anaemia, Plummer-Vinson syndrome, megaloblastic anaemia, sickle cell anaemia, thalassemia and aplastic anaemia.

TREATMENT

Only symptomatic treatment offered as no specific protocols was available. The goal of treatment is to manage symptoms. Folic acid supplements were administered to help in haemopoiesis.

Antibiotic (amoxycillin 500 mg 8th hourly perorally for 7 days) was administered as bacterial infections are common in children with sickle cell disease.

Figure 1 Showing swelling in right infraorbital region.

Figure 2 Showing swelling in left cheek.

DISCUSSION

Normocytic haemolytic anaemia is a feature of sickle cell disease. As a result of polymerisation and the subsequent damage to red blood cell (RBC) membranes during oxygenation and deoxygenation, approximately one-third of RBCs undergo intravascular haemolysis. Chronic anaemia leads to an increased need for blood production and results in intra-medullary or extra-medullary haematopoiesis.¹⁻³ Because of this increased need for blood, chronic anaemia is associated with increased cardiac output and high blood flow velocity, and it may lead to arterial tortuosity.⁴

Orbital wall infarction is an uncommon manifestation of sickle cell disease; however, in the literature, there are many isolated case reports of patients who presented with acute, rapidly progressive periorbital pain and swelling, manifestations indicative of infarction.⁵⁻¹⁰ Orbital wall infarction typically occurs in youths because there is more marrow space in the orbital bone in children than there is in adults.⁵ A unique feature of orbital wall

Figure 3 Orthopantomogram showing no evidence of dental origin.

infarction in sickle cell disease is the formation of haematomas, which may be orbital (subperiosteal) or intracranial (epidural).⁵⁻¹⁰ Vaso-occlusion of orbital bone marrow leads to orbital wall infarction. Several mechanisms have been suggested to cause subperiosteal haemorrhage, such as extravasation of blood from necrosed vessel walls, underlying bleeding diathesis and minor trauma.⁶⁻⁹ This could explain the sudden onset swellings seen in the case presented.

Extramedullary haematopoiesis occurs in chronic anaemic conditions such as thalassaemia, sickle cell disease and myeloproliferative disorders.¹⁻³ It is hypothesised that extramedullary haematopoiesis is a compensatory response to an increased need for blood production. It most commonly occurs in the liver and spleen; however, it also is associated with the paravertebral regions, kidneys and adrenal glands.¹¹ Although it is rare in the head and neck, it has been reported in the paranasal sinuses, middle ear, thyroid gland, cervical lymph nodes and lacrimal fossa.¹⁻³

In the paranasal sinuses, extramedullary haematopoiesis has been reported in the maxillary, ethmoid and sphenoid sinuses; the maxillary sinus is most commonly involved.¹⁻³ Extramedullary haematopoiesis is a result of herniation of medullary tissue into the paranasal cavity due to underlying bone marrow hyperplasia.¹²

Bone involvement is the most common clinical manifestation of sickle cell disease.¹³⁻¹⁴ However, it is rare in the maxillofacial bone and skull base because of the small amount of marrow space within these bones. In the head and neck, the most frequently reported location is the orbital wall, followed by the mandible and skull base. There are acute and chronic phases of bone involvement. In the acute phase, vaso-occlusive crisis and osteomyelitis are seen. In the chronic phase, bone marrow hyperplasia, osteoporosis and iron deposition in marrow because of repeated transfusions may be seen.¹⁴

Vaso-occlusive crisis is about 50 times more common than osteomyelitis among children with sickle cell disease.¹³ In the first few days of a vaso-occlusive crisis, abnormal signal intensity in the periosteum and soft-tissue changes are seen, with severe medullary involvement and compromised periosteal circulation. After a vaso-occlusive crisis occurs in the mandible, permanent neuropathies affecting the inferior alveolar nerve, which can result in persistent anaesthesia for up to 24 months, have been reported.

Lymph node swelling is a common finding in patients with sickle cell disease, but it is non-specific. When lymph node swelling is present, lymphadenopathy should be suspected, because patients with sickle cell disease are at increased risk for infection. When lymph nodes are involved, CT and MRI depict bilateral cervical lymphadenopathy. Patients with sickle cell disease have been reported to develop haematological malignancies such as lymphoma, multiple myelomas and hairy cell leukaemia, although the relationship between these malignancies and sickle cell disease has not been clearly defined.

Cerebrovascular involvement leads to brain infarction, one of the most severe and most common complications of sickle cell disease, affecting 25% of patients with the disease.¹³ Vascular involvement is classified as arterial tortuosity, stenosis, occlusion or aneurysm, according to its imaging appearance. Tortuosity is subjective and is

determined on the basis of the following features: (1) dilatation (ectasia) of a vessel segment, (2) abnormal increase in length of a vessel segment and (3) obvious bowing of an artery. Arterial tortuosity is considered to be an adaptive response to chronic anaemia in patients with sickle cell disease. Because chronic anaemia is associated with increased cardiac output and high blood flow velocity due to the increased need for blood supply, it can lead to arterial tortuosity.⁴

Learning points

- A wide array of disorders of red cells and haemostasis encountered in internal medicine has manifestations in the oral cavity and the facial region. Most of these manifestations are non-specific, but should alert the haematologist and the dental surgeon to the possibilities of a concurrent disease of haemopoiesis or haemostasis or a latent one that may subsequently manifest itself. These manifestations must be properly recognised if the patient must receive appropriate diagnosis and referral for treatment. Proper diagnosis is essential to initiate the correct treatment.
- Sickle cell disease is a common inherited blood disorder that can involve virtually any organ system. Head-and-neck involvement is not uncommon, although it often is underappreciated.
- The clinical and radiological manifestations of sickle cell disease in the head and neck are the same as those of systemic involvement and may be attributed to vaso-occlusion, chronic anaemia or infection. Familiarity with these manifestations is crucial to provide an accurate, timely diagnosis and optimal treatment.

Competing interests None.

Patient consent Obtained.

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