International Journal of Pediatric Otorhinolaryngology 76 (2012) 1–4

Contents lists available at SciVerse ScienceDirect

International Journal of Pediatric Otorhinolaryngology

journal homepage: www.elsevier.com/locate/ijporl

Review article

Otorhinolaryngological manifestations of sickle cell disease

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ARTICLE INFO

Article history:
Received 31 August 2011
Received in revised form 3 October 2011
Accepted 4 October 2011
Available online 22 October 2011

Keywords:
Sickle cell disease
Otorhinolaryngology
Extramedullary hematopoiesis
Adenotonsillar hypertrophy
Sensorineural hearing loss
Priapism

ABSTRACT

Objectives: Although sickle cell disease is the most common of all hereditary disorders, there are a few publications about the effects of the disease on the functions of the ears, nose and throat.

In this review, we present an overview of the clinical manifestations of sickle cell disease in general and highlight the problems specifically presenting in the field of otorhinolaryngology.

Methods: We review the pathophysiology of sickle cell disease and its clinical features in general. Then, we review its manifestations in ear, nose and throat diseases.

Conclusion: Sickle cell disease is one of the most common hemoglobinopathies. It can cause severe pain crises and dysfunction of virtually every organ system in the body, ultimately causing premature death. There is high prevalence (55%) of obstructive adenotonsillar hypertrophy in children and adolescents with sickle cell disease. A very significant reduction has been observed in the rate of pain crises following tonsillectomy in patients with sickle cell anaemia. Prevalence rate for sensorineural hearing loss in older children and adult patients is reporting a range of 11–41%. Priapism of the turbinates is a cause of nasal obstruction in sickle cell anaemia which needs partial turbinectomy. Extramedullary haematopoiesis should be considered in the differential diagnosis of any paranasal sinus mass presenting in a patient with known chronic anaemia.

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doi:10.1016/j.ijporl.2011.10.004

1. Introduction

1.1. Pathophysiology

1.1.1. What is sickle cell?

Sickle cell is the red blood cell that changes from normal biconcave disc shaped to sickle shaped. Sickle cells die faster than normal red blood cells, usually after only about 10–20 days. The bone marrow cannot make new red blood cells fast enough to replace the dying ones. The result is anaemia. Sickle-shaped cells do not move easily through blood. They are stiff and sticky and tend to form clumps and get stuck in blood vessel.

1.1.2. What is sickle cell disease?

Sickle cell disease (SCD) was first described by Herrick in 1910 [1]. It is a group of hemoglobin disorders in which the sickle β-globin gene (a variant of the β-globin gene on chromosome 11) is inherited as an autosomal recessive disorder. A glutamic acid at the 6th position is replaced by a valine resulting in formation of Hemoglobin S (Hb S). Hb A has 4 protein subunits (α_2 β_2). There will be different forms of hemoglobin when there is a mutation in the beta subunit.

1.1.3. What is sickle cell anaemia?

It is the homozygous state which occurs when both genes are abnormal – S mutation (Hb SS). It is the most common and most severe form. Both copies of the hemoglobin beta gene have S mutation. All beta subunits are replaced by beta S. Fetal hemoglobin (Hb F) normally accounts for around 80% of the hemoglobin concentration at birth, usually reducing to less than 1% by the age of 6 months and gives protection against the complications of SCD over the first few months of life.

1.1.4. What is sickle cell trait?

It is the heterozygous state which occurs when only one chromosome carries the gene (Hb AS). A carrier for sickle cell will have an S mutation in one copy of the hemoglobin beta gene. Half of the beta subunits are replaced with Beta S.

1.1.5. Genetics of sickle cell

Sickle cell is an autosomal recessive disease. Therefore, the child can only get sickle cell if both parents are carriers. They have a 25% chance of getting it if both are carriers.

1.1.6. Distribution

The prevalence of SCD is high among Africans and their descendents, Arabs, Central and South Americans, West Indies and people of the Mediterranean countries, such as Turkey, Greece, and Italy [1].

Sickle cell anaemia affects up to 8% of African Americans. The majority of these patients have sickle cell trait. Sickle cell disease affects approximately 0.2% of African Americans. The prevalence of sickle cell disease in non-African Americans is approximately 0.08% [2].

1.1.7. In Saudi Arabia

In the Eastern Province, 20–30% of Saudi newborns are heterozygous for the sickle gene and 1.6–2.3% may be homozygous [3]. Two well-defined forms of SCD are seen in Saudis. In the eastern province, the disease is milder, while in the western province the disease is severe [4]. Patients from the eastern part of Saudi Arabia carry the Saudi-Indian b-globin gene haplotype, while the Benin haplotype is the major one in the western areas of Saudi Arabia, Egypt, Jordan, and Syria [5].

1.2. Clinical features

It is characterized by frequent episodes of hemolysis in the form of four types of crises. Sickle cell crisis can last for hours or weeks and may occur several times per year. The cardinal symptoms of sickle cell crisis are severe pain, fever, oedema and inflammation. Generally the patient is in fatigue, pale and has jaundice and complains of dyspnea.

The crises are painful vaso-occlusive crises, visceral sequestration crises, aplastic crises and hemolytic crises.

1.2.1. Painful vaso-occlusive crises

It is the most frequent crises and it is precipitated by infections, acidosis, dehydration, or deoxygenation (e.g. altitude, operations, obstetric delivery, and exposure to cold, violent exercise....). Emotional stress, physical exertion, and alcohol consumption may also precipitate the crises. Infarcts may occur in variety of organs such as bones (hips, shoulders, vertebrae), lungs, spleen and brain and spinal cord (most serious). Hand-foot' syndrome is common in these patients which is painful dactylitis caused by infarcts of the small bones owing to vaso-occlusive of small vessels.

1.2.2. Visceral sequestration crises

Dyspnea, falling PO_2, chest pain, splenic sequestration are typically seen in infants and young children due to visceral crises. In puberty, sickling within the organs and acute chest syndrome are common and lead to death. Pooling of RBCs and hypovolemia as a result of visceral crises can also lead to circulatory collapse and death.

1.2.3. Aplastic crises

It results from infection with parvovirus or folate deficiency leading to sudden fall in hemoglobin and usually requires transfusion. Fall in reticulocytes (failure of erythropoiesis) is a usual finding.

1.2.4. Hemolytic crises

Increase rate of hemolysis results in fall in hemoglobin and rises in reticulocytes.

1.2.5. Other clinical pictures

Patients may have ulcers of lower legs due to vascular stasis and local ischemia. Splenomagally in infants and young children are common. Later the spleen reduced in size as a result of infarction (autosplenectomy). Proliferative retinopathy, priapism and pigment gallstones are frequent. Kidneys infection may happen. Osteomylitis is frequent and it usually is due to Salmonella infection.

However, the presence of sickle trait in individuals provided natural protection against malaria.