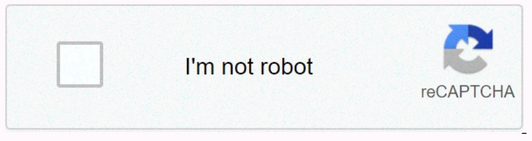


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Mansour, Sohier Yahia, Rasha El-Ashry, Angi Alwakeel, Ahmad Darwish and Khalil AlrjjalPreliant: November 13, 2014Re view: July 3, 2015Published: November 11, 2015DOI: 10.5772/61162Sickle cell anaemia (SCA) is a disease caused by the formation of an abnormal type of haemoglobin that can bind to other abnormal molecules of haemoglobin in red blood cells (RBC) to cause firm cell distortion I don't know This distortion prevents the cell from passing through small blood vessels; leads to occlusion of the vascular beds, followed by tissue ischemy and heart attack. Heart attack is common throughout the body in patients with SCA, which leads to an acute pain crisis. Over time, these insults result in medullary bone infarctions and epiphysical osteonecrosis. In the brain, cognitive disorders and functional neurological deficits can occur due to white matter and gray matter heart attacks. A heart attack can also affect the lungs increasing susceptibility to pneumonia. The liver, spleen and kidneys may show a heart attack as well. Sequestration crisis is an unusual life-threatening complication of SCA, in which a significant amount of blood is sequestered in the organ (usually the spleen), leading to collapse. Finally, because RBCs are abnormal, they are destroyed, resulting in hemolytic anemia. However, ischaemic complications in patients with SCA go far beyond anaemia of clinical relevance. Hemoglobin is necessary for the transfer of oxygen to various body organs. The shape of red blood cells may be affected by the type of haemoglobin. Hemoglobinopathy are abnormalities of hemoglobin that affect its formation. The severity of these disorders varies greatly and can lead to death. Hemolytic anemia is a common presentation of hemoglobinopathy. Sickle cell anemia is one of these hemoglobinopathy. Sickle cell anaemia is an inherited disease characterised by the presence of an abnormal haemoglobin called haemoglobin S (HbS). During the shape of red blood cells (RBC) varies from biconcave to sickle cell due to abnormal haemoglobin. The RBC shape changes back to a biconcavular shape after reoxygenation. However, frequent sick and unwavering leads to hemolysis and anemia. [1] There are three types of normal haemoglobin: haemoglobin A (HbA), haemoglobin F (HbF) and haemoglobin A2 (HbA2). Each hemoglobin molecule contains four polypeptide chains that vary from one type to another. Haemoglobin A contains 2 alpha globin chains and two beta globin chains and contains 95-97% of normal haemoglobin. Haemoglobin A2 contains 2 alpha globin chains and two gamma globin chains and contains 2.5-3.5% of normal haemoglobin. Hemoglobin F contains 2 alpha globin chains and two delta globin chains and <1% normal haemoglobin. Gene coding for α globin chain is located on chromosome 16, however, the α globin gene cluster is found on chromosome 11. [2,3] In the sixth cone of the globin gene β from A to T produced by HbS, the mutation is in the sixth position of the amino acids in the globin polypeptide chain β, which is to be a valine instead of glutamic acid. Patients with sickle cell anaemia (homozygous to HbS gene) have HbS instead of HbA associated with hbf and HbA2 production. Some patients with sickle cell anaemia (double heterozygous) received HbS along with other types of abnormal haemoglobin or even sickle cell-thalassaemia. However, thalassemias on their own occur more often, leading to homozygous disease conditions. [4] Abnormal haemoglobin is responsible for haemolysis and vaso-occlusion, which can lead to tissue infarction. [5,6] Abnormalities of hemoglobin and thalassemia are hereditary as autosomal recessive (AR) disorders, where carrier parents transmit the disease to their offspring. If both parents are heterozygotes for HbS, there is a 25 percent chance of having a homozygous HbSS (sickle cell anemia, SCA) child. A double heterozygous state occurs when one parent is a heterozygote for HbS and the other is heterozygous for one of the abnormal HbS or thalassemia. Heterozygotes are asymptomatic carriers (properties), while SCD is given in homozygotes and double heterozygots for two abnormal hemoglobin or HbS and thalassemia genes. [6] Sickle cell anaemia is a single gene disorder produced by a point mutation in the beta globin gene located on chromosome 11. This leads to the replacement of glutamic acid (hydrophilic amino acid) in the sixth position with a valine (hydrophobic amino acid). [7] Haemoglobin S is made up of an association α-globin sububine units with two mutant β-globin subutech units. When exposed to hypoxic conditions, the absence of RBCs.In a polar amino acid at position six of the β-globin chain promotes necilly polymerisation (aggregation) of hemoglobin, which changes the shape and elasticity of the low-acid medium, the cells achieve an abnormal which is not flexible. When the normal oxygen voltage is regained, the cells do not return to normal shape. Therefore, these distorted RBCs can not pass through narrow capillaries, which leads to occlusion of blood vessels. Vasooklusion results in hand-foot syndrome in children. In addition, infections, strokes, and acute chest pain are some of the main complications. Most of these complications begin at an early age, but more clearly with advancing age. Infections, dehydration, cold weather and stress are considered precipitating factors for these complications. Treatment of SCD are mostly aimed at preventing or reducing sickle, thereby reducing the incidence of vascular occúsia. [5-10] Abnormal RBC shape leads to their destruction by haemolysis. Bone marrow compensation hyperplasia is not able to match the rate of RBC destruction. [8] Sickles survive only 10 to 20 days compared to normal ERYTHROMYtes, which usually live for 90-120 days. [9] Sickle cell disease is most common among people from Africa, India, the Caribbean, the Middle East and the Mediterranean. In the Middle East came the first report on HbS and thalassemias from Egypt. [11.12] The presence of HbS in eastern Saudi Arabia has been reported by Lehmann. [13] Many studies on haemoglobinopathy have been documented from most Middle Eastern countries. Table (1) presents a brief history for identifying abnormal haemoglobins in the Middle East. HbS is the main variant identified in all areas. [14] Discovery countryYearFirst case of SCD in Egypt1951HbS in Middle East1959HbO-Arab in Egyptian family1960HbS in Saudi Arabia1963HbS and HbO-Arab in Sudan1966HbC in Egyptians1967Mild SCD in Saudi Arabia1969SCD in Kuwait19 69HbH disease in Kuwait1969HbS in Egypt's western desert1974HbC in Libya1975HbS in Abu Dhabi1980HbC in Saudi Arabia1979HbE and HbD in Abu Dhabi1979HbO-arab in Saudi Arabia1980HbS. α- and β-thal in several regions of Saudi Arabia1967-1982Hemoglobinopathies in the Middle Eastern Arab countriesAphtic cell anemia represents severe hemolytic anemia interrupted by the crisis. Symptoms of anemia in SCD are often mild in relation to the severity of anemia because HbS gives up oxygen (O2) to tissues relatively easily compared to HbA, its O2 dissociation curve shifts to the right (see Figure 1). [15] Oxygen dissociation curve of haemoglobin. 2,3-DPG, 2,3-dipphosphoglycerate. Clinical presentation of SCD is variable, in some patients with normal life; However, some patients show increased morbidity and mortality due to severe thrombotic, aplastic and sequestration crises. [15] These crises are caused by pooling blood, with severe exacerbation of anemia. Acute sickle cell syndrome is the most common cause of death after puberty. Patients present with dyspnea, arterial hypoxia, chest pain, and pulmonary infiltrates on the chest X-ray. Treatment includes analgesics, oxygen, transfusion replacement, and fan support if necessary. Liver and sequestration may lead to a serious disease requiring the exchange of transfusions. Sequestration of the spleen is characteristically found in infants and is clinically manifested with increasing spleen, decreased hemoglobin and abdominal pain. Patients are mainly treated with a blood transfusion and should be monitored frequently as rapid progression may occur. Crises are usually recurrent and the patient is usually in need of a splenectomy. [15] Aplastic crises are caused by parvovirus infection and are characterized by a sudden decrease in hemoglobin, which usually requires transfusion. The patient exhibits anemia along with reticulocytopenia. [15] In these crises, patients show higher haemolysis rates with a decrease in haemoglobin associated with reticulocytosis. Hemolytic crises usually accompany a vaso-occlusive crisis. Chronic haemolytic anaemia is the main clinical presentation of SCDs with recurrent bouts of acute painful vaso-occlusive crises. SCD is also associated with multi-organ acute and chronic complications. The clinical characteristics of SCD are summarised in Table (2). The size of the spleen increases during childhood and early childhood, but later it usually decreases due to a heart attack (autosplenectomy). Pulmonary hypertension and tricuspid regurgitation can occur and increase the risk of mortality. Retinopathy and priapism can also complicate the course of patients with SCD. Chronic liver damage can occur due to microinfarction associated with gallbladder stones. Renal meduary infarction with papillary necrosis may be present during sickle cell anemia. The ability of the kidneys to concentrate urine can be lost, which leads to dehydration and vaso-occlusive crises, and night-time enuresis is common. [15] Although genetic variation in SCD is well understood, there is a clear variability in the clinical severity of the disease among patients. Some patients lead normal lives, without problems; severe crises or have fatal complications. The life expectancy of PATIENTS with SCD decreases, but increases due to improved supportive therapies, in particular prophylactic antibiotics, early childhood stroke screening, increased hydroxycarbamide or transfusion administration, and better care. Intensive care is needed in patients complicated by acute chest disease (ACS), acute stroke or acute renal injury. [16] ComplicationsClinical presentationsAdest crisis These crises occur in most patients with SCD; are variable in frequency and severityYou can lead to chronic pain syndrome Neurological ocular occliology can be seen on mri. It can lead to cognitive impairment. Stroke affects 10% of children; is the main cause of morbidity and mortality. Can be prevented by regular blood transfusionPulmonaryAkut chest syndromeAsthma, fibrotic lung diseaseThe main cause of death in adults, high risk of acute respiratory failure There is an increased association with the airways HepatopathyMome of patients have gallstones due to hemolysisCompensated liver disease may be present in some patientsRenal, UrologicalChronic renal failure occurs in 20% of patientsPriapism may be present leading to sexual dysfunctionOftalmologyProliferative retinopathy is common in patients with HbSCOrthopedicAccular necrosisOsteomyelitisCometic complication of the hip and shoulders, requiring replacementSalmonella is the most common organismHematological haemolytic anemiaAplastic crisisSlezinic sequestrationChronic hemolysis, hb 6-9 g/dL, higher in HbSCParvovir B19 infection may cause red blood cell alasia. The combination of red blood cell aplasia and haemolysis can be fatalTypically seen in infants with rapidly increasing spleenClinical presentation of sickle cell anaemia (SCD)In HbSS, the full blood count shows haemoglobin levels in the range of 6-8 g/ol with reticulocytosis (due to compensatory bone marrow hyperplasia). With other forms of sickle cell anaemia, Hb levels tend to be higher. A blood film can reveal sickle cell and signs of hyposplenism (target cells and Howell-Jolly's bodies) (Figure 5). [15] Sickle cell anaemia: peripheral blood foil showing deep staining of sickle cell, target cells and polychromasia. Hemoglobin electrophoresis is used to diagnose the presence of abnormal types of hemoglobin. Hemoglobin S and hemoglobin SC are the two most common forms detected in patients sick with sickle cell anemia. High-end liquid chromatography (HPLC) is used to confirm the diagnosis. Genetic studies are often not conducted because electrophoresis and HPLC are accurate in detecting HbS and HbC.[17]Infection can accelerate an acute sickle cell crisis. Therefore, urine analysis should be performed to detect occult urinary tract infection and chest X-ray to detect occult pneumonia. [18] Genetic counselling is usually required for SCD carriers before they have a child. Fetal blood collection or amniocentesis can be performed to see if the fetus has the disease. Miscarriage is more common when taking the blood of the fetus than with amniocentesis. Treatment of crises is usually supportive unless blood transfusion is indicated. The goal of treatment is to prevent erythrocyte sickle, dehydration, hypoxia and acidosis, which can provoke sickle. A painful attack is the main presentation. Subcutaneous morphine or another powerful opioid is often needed to manage severe bouts of pain. Peticin can speed up Grand mal seizures; it is therefore preferable to avoid it. Satisfactory fluid intake is mandatory. Children born with sickle cell anemia will take folic acid (1 mg dose) a day for life. In addition, patients from birth to five years of age must take penicillin daily due to susceptibility to pneumococcal infection. Acute thoratic syndrome is an acute disease with fever and/ or respiratory symptoms associated with a new pulmonary infiltrate. It is the leading cause of mortality in adults with SCD and the most common cause of Adoption. The patient, who needs mechanical ventilation, is reported to have a mortality rate of 5%. [16] Symptoms include coughing, wheezing, dyspnoea and chest pain, which may be pleurotic or affect the ribs and sternum. Acute thoratic syndrome is unique to SCD and is associated with a more severe course and worse outcome than pneumonia. Blood transfusion is used to treat patients with acute thoratic syndrome and improve oxygenation. Blood transfusion is useful in less severe cases with low Hb (<7 g/dL); however, in severe cases, foreign exchange transfusion is required in patients with high Hb levels or in patients with severe hypoxia. The goal is a final level of Hb 9–10 g/0L. Severe hypoxia, dyspnoea and respiratory acidosis are indications for initiating advanced respiratory support. [19] Patients with SCD are commonly associated with ischaemic and haemorrhagic stroke, with a prevalence rate of more than 5%. The incidence of stroke decreases significantly after the introduction of transcranal Doppler screening and primary prevention of transfusion stroke. Stroke can be precipitated by dehydration or coincidentally disease. Timely imaging is necessary to confirm the diagnosis and exclude bleeding. MRI is a display of choice with high sensitivity and specificity. If an MRI confirms a stroke, an immediate foreign exchange transfusion should be performed to achieve HbS of less than 30%. Ischaemic prevention of stroke may be performed by long-term exchange transfusion, but the efficacy of anti-platelet therapy in primary or secondary prevention of stroke in SCD is not demonstrated. [20] Patients with sickle cell anaemia have functional hyposplenism. This makes them more susceptible to infection by capulated organisms. Sepsis caused by Gram-negative organisms is common along with osteomyelitis. Children with sickle cell anaemia must be vaccinated against pneumococcal, meningococcal and haemophilic influenza infection. Oral penicillin can be administered daily after diagnosis to protect against pneumococcal infection. [21] Patients with SCD have a low ability to concentrate on the kidneys and are therefore prone to dehydration. Over time, patients may experience proteinuria and chronic kidney damage due to glomerular damage. This leaves patients prone to acute kidney injury during the crisis. Chronic lung disease is common and manifests itself either as a restrictive lung defect or overnight hypoxia and sleep apnea. Pulmonary hypertension is more common in SCD and can lead to severe hypoxia. [16] Patients with sickle cell anaemia may need admission to the intensive care unit either due to liver cell failure, sepsis or multiple organ damage. This acute deterioration may require an urgent blood transfusion targeting Hb 9-10 g/0L and HbS% less than 30%. This will improve tissue oxygenation and perfusion, regardless of basic etiology. [22] Regular blood transfusion is necessary in order to prevent a stroke. Specific situations such as circulatory system failures, sequesterary crisis crises môže potrebovať transfúziu krvi na optimalizáciu transportu kyslíka. [23] Čiastočná výmenná transfúzia sa zvyčajne uprednostňuje pred jednouchou transfúziou, ak sú potrebné rutinné alebo viacnásobné transfúzie. Znižuje preťaženie železom a zabraňuje zvýšenej viskozite krvi. Existujú určité línie liečby, ktoré znižujú chorobnosť a úmrtnosť u detí s kosáčikovitou anémiou, vrátane:Očkovanie proti kapulovaným organizmom (napr. Hydroxyurea a kyselina listová suplementácia. Perorálna profylaxia penicilínu u detí mladších ako 6 rokov. Včasná detekcia a riadenie závažných bakteriálnych infekcií. Hydroxyurea zvýšením HbF, a tým znížením kosáka, znižuje bolestivé krízy (o 50%) a znižuje akútny hrudný syndróm a transfúzne požiadavky. Dávka hydroxyurea je variabilná a upravuje sa tak, aby sa zvýšila HbF.Hydroxyurea je účinnejšia u niektorých pacientov, ak sa podáva s erytropoetínom (40 000 - 60 000 jednotiek/týždeň). Hydroxyurea však môže spôsobiť neutropéniu a trombocytopéniu. Hydroxyurea je tiež teratogén a nemá sa podávať samici v období loženia dieťaťa. Skríning cievnej mozgovej príhody u detí s SCD sa odporúča vykonať vo veku od 2 do 16 rokov pomocou transkraniálnej Štúdie toku Doppler. Risky children can get benefit from prophylactic, chronic partial exchange transfusions keeping HbS at < 30% of total Hb.Erythropoietin use in patients with anemia not related to chemotherapy is associated with high incidence of venous thromboembolism and cardiopulmonary complications (as myocardial infarction); it is not useful in patients with sickle cell disease except possibly if given in combination with hydroxyurea. [23] Omega-3 fatty acids are significantly reduced in SCD patients. In a single-center study conducted in Sudan, there was a randomized, placebo-controlled, double-blind design for studying the effect of omega-3 treatment on sickle cell anemia patients. One hundred and forty patients were monitored for 1 year, and it was found that omega-3 treatment leads to a decline in occlusive crises and blood transfusion. Treatment with omega-3 was well tolerated by the patients and needs further study. [24] It is a new thienopyridine P2Y12 ADP receptor antagonist, which inhibits ADP-mediated platelet activation and aggregation. Phase 2 randomized, double-blind, placebo controlled studies to examine safety were completed in adults. There were no hemorrhagic events requiring medical intervention in either study arm. Mean pain rates (percentage of days with pain) and intensity in the prasugrel arm were decreased compared with placebo. But, these results were not statistically significant. It was well tolerated and a phase 3 trial in children is registered. [25] The life span of homozygous patients with SCD has gradually increased to > 50 years. Častým príčinám smrti sú akútny hrudný syndróm, opakujúce sa infekcie, pľúcna embólia, infarkt životne dôležitého orgánu a zlyhanie obličiek. [23] Kosáčikovkej anémie je haemoglobinopathy mainly affecting blacks and leading to chronic haemolysis. Abnormal HbS found in homozygous patients changes the shape of Erythrocyte kits to become sickle cell. These cells can occute small blood vessels leading to ischemy and pain. Patients can be complicated by acute thoratic syndrome, sepsis, sequestration and aplastic crises. Sickle cell anemia is characterized by anemia and can be diagnosed with HB electrophoresis. Blood transfusions may be required for these patients. Occual crises are mainly treated with painkillers. Hydroxyurea can reduce the frequency of these crises. Early management of bacterial infections and vaccination against capulated organisms can prevent sepsis.1541Some IntechOpen, the world's leading publisher of Open Access books. Built by scientists, for scientists. Our reading activities include scientists, professors, researchers, librarians and students, as well as business professionals. 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