DOWNLOAD PDF THE CLINICAL FEATURES OF SICKLE CELL DISEASE

Chapter 1 : Sickle Cell Disease | National Heart, Lung, and Blood Institute (NHLBI)

The clinical spectrum of homozygous sickle cell disease varies widely between patients. Factors contributing to this variability include alpha-thalassaemia, persistence of high HbF levels, haematology, social circumstances, and geographical and climatic variation.

Sickle cell anemia Sickle cell anemia Normal red blood cells are round. In sickle cell anemia, some red blood cells become deformed, so they look like sickles used to cut wheat. These unusually shaped cells give the disease its name. Normally, your red blood cells are flexible and round, moving easily through your blood vessels. In sickle cell anemia, the red blood cells become rigid and sticky and are shaped like sickles or crescent moons. These irregularly shaped cells can get stuck in small blood vessels, which can slow or block blood flow and oxygen to parts of the body. But treatments can relieve pain and help prevent problems associated with the disease. Symptoms Signs and symptoms of sickle cell anemia, which vary from person to person and change over time, include: Sickle cells break apart easily and die, leaving you without enough red blood cells. Red blood cells usually live for about days before they need to be replaced. But sickle cells usually die in 10 to 20 days, leaving a shortage of red blood cells anemia. Periodic episodes of pain, called crises, are a major symptom of sickle cell anemia. Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to your chest, abdomen and joints. Pain can also occur in your bones. The pain varies in intensity and can last for a few hours to a few weeks. Some people have only a few pain episodes. Others have a dozen or more crises a year. If a crisis is severe enough, you might need to be hospitalized. Some adolescents and adults with sickle cell anemia also have chronic pain, which can result from bone and joint damage, ulcers and other causes. Painful swelling of hands and feet. The swelling is caused by sickle-shaped red blood cells blocking blood flow to the hands and feet. Sickle cells can damage an organ that fights infection spleen , leaving you more vulnerable to infections. Doctors commonly give infants and children with sickle cell anemia vaccinations and antibiotics to prevent potentially life-threatening infections, such as pneumonia. Red blood cells provide your body with the oxygen and nutrients you need for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers. Tiny blood vessels that supply your eyes may become plugged with sickle cells. This can damage the retina â€” the portion of the eye that processes visual images, leading to vision problems. When to see a doctor Although sickle cell anemia is usually diagnosed in infancy, if you or your child develops any of the following problems, see your doctor right away or seek emergency medical care: Unexplained episodes of severe pain, such as pain in the abdomen, chest, bones or joints. Swelling in the hands or feet. Abdominal swelling, especially if the area is tender to the touch. People with sickle cell anemia have an increased risk of infection, and fever can be the first sign of an infection. Pale skin or nail beds. Yellow tint to the skin or whites of the eyes. Signs or symptoms of stroke. If you notice one-sided paralysis or weakness in the face, arms or legs; confusion; trouble walking or talking; sudden vision problems or unexplained numbness; or a headache, call or your local emergency number right away. Request an Appointment at Mayo Clinic Causes Sickle cell anemia is caused by a mutation in the gene that tells your body to make the red, iron-rich compound that gives blood its red color hemoglobin. Hemoglobin allows red blood cells to carry oxygen from your lungs to all parts of your body. In sickle cell anemia, the abnormal hemoglobin causes red blood cells to become rigid, sticky and misshapen. The sickle cell gene is passed from generation to generation in a pattern of inheritance called autosomal recessive inheritance. This means that both the mother and the father must pass on the defective form of the gene for a child to be affected. If only one parent passes the sickle cell gene to the child, that child will have the sickle cell trait. With one normal hemoglobin gene and one defective form of the gene, people with the sickle cell trait make both normal hemoglobin and sickle cell hemoglobin. But they are carriers of the disease, which means they can pass the gene to their children. Risk factors For a baby to be born with sickle cell anemia, both parents must carry a sickle cell gene. In the United States, it most commonly affects black people. Complications Sickle cell anemia can lead to a host of complications, including: A stroke can occur if sickle cells block blood flow to an area of your brain. Signs of stroke include

seizures, weakness or numbness of your arms and legs, sudden speech difficulties, and loss of consciousness. If your baby or child has any of these signs and symptoms, seek medical treatment immediately. A stroke can be fatal. This life-threatening complication causes chest pain, fever and difficulty breathing. Acute chest syndrome can be caused by a lung infection or by sickle cells blocking blood vessels in your lungs. It might require emergency medical treatment with antibiotics and other treatments. People with sickle cell anemia can develop high blood pressure in their lungs pulmonary hypertension. This complication usually affects adults rather than children. Shortness of breath and fatigue are common symptoms of this condition, which can be fatal. Sickle cells that block blood flow through blood vessels immediately deprive the affected organ of blood and oxygen. In sickle cell anemia, blood is also chronically low on oxygen. Chronic deprivation of oxygen-rich blood can damage nerves and organs in your body, including your kidneys, liver and spleen. Organ damage can be fatal. Sickle cells can block tiny blood vessels that supply your eyes. Over time, this can damage the portion of the eye that processes visual images retina and lead to blindness. Sickle cell anemia can cause open sores, called ulcers, on your legs. The breakdown of red blood cells produces a substance called bilirubin. A high level of bilirubin in your body can lead to gallstones. Men with sickle cell anemia can have painful, long-lasting erections, a condition called priapism. As occurs in other parts of the body, sickle cells can block the blood vessels in the penis. This can damage the penis and lead to impotence. Prevention If you carry the sickle cell trait, seeing a genetic counselor before trying to conceive can help you understand your risk of having a child with sickle cell anemia. He or she can also explain possible treatments, preventive measures and reproductive options.

Chapter 2 : Sickle cell disease - Wikipedia

Despite the title, the book is not limited strictly to the clinical features of sickle cell disease; the historical background, biochemical features, diagnostic tests, and worldwide geographical distribution of the sickle hemoglobin are initially presented.

The following are types of complications that can result from sickle cell anemia. Severe anemia Anemia is a shortage of RBCs. Sickle cells are easily broken. This breaking apart of RBCs is called chronic hemolysis. RBCs generally live for about days. Sickle cells live for a maximum of 10 to 20 days. Hand-foot syndrome Hand-foot syndrome occurs when sickle-shaped RBCs block blood vessels in the hands or feet. This causes the hands and feet to swell. It can also cause leg ulcers. Swollen hands and feet are often the first sign of sickle cell anemia in babies. Splenic sequestration Splenic sequestration is a blockage of the splenic vessels by sickle cells. It causes a sudden, painful enlargement of the spleen. The spleen may have to be removed due to complications of sickle cell disease in an operation known as a splenectomy. Some sickle cell patients will sustain enough damage to their spleen that it becomes shrunken and ceases to function at all. This is called autosplenectomy. Patients without a spleen are at higher risk for infections from bacteria such as Streptococcus, Haemophilus, and Salmonella species. Delayed growth Delayed growth often occurs in people with SCD. Children are generally shorter but regain their height by adulthood. Sexual maturation may also be delayed. Neurological complications Seizures, strokes, or even coma can result from sickle cell disease. They are caused by brain blockages. Immediate treatment should be sought. Eye problems Blindness is caused by blockages in the vessels supplying the eyes. This can damage the retina. Skin ulcers Skin ulcers in the legs can occur if small vessels there are blocked. Heart disease and chest syndrome Since SCD interferes with blood oxygen supply, it can also cause heart problems which can lead to heart attacks , heart failure , and abnormal heart rhythms. Lung disease Damage to the lungs over time related to decreased blood flow can result in high blood pressure in the lungs pulmonary hypertension and scarring of the lungs pulmonary fibrosis. These problems can occur sooner in patients who have sickle chest syndrome. Lung damage makes it more difficult for the lungs to transfer oxygen into the blood, which can result in more frequent sickle cell crises. Priapism Priapism is a lingering, painful erection that can be seen in some men with sickle cell disease. This happens when the blood vessels in the penis are blocked. It can lead to impotence if left untreated. Gallstones Gallstones are one complication not caused by a vessel blockage. Instead, they are caused by the breakdown of RBCs. A byproduct of this breakdown is bilirubin. High levels of bilirubin can lead to gallstones. These are also called pigment stones. Sickle chest syndrome Sickle chest syndrome is a severe type of sickle cell crisis. It causes severe chest pain and is associated with symptoms such as cough, fever, sputum production, shortness of breath, and low blood oxygen levels. Abnormalities observed on chest X-rays can represent either pneumonia or death of lung tissue pulmonary infarction. The long-term prognosis for patients who have had sickle chest syndrome is worse than for those who have not had it. How is sickle cell anemia diagnosed? All newborns in the United States are screened for sickle cell disease. Prebirth testing looks for the sickle cell gene in your amniotic fluid. In children and adults, one or more of the following procedures may also be used to diagnose sickle cell disease. Detailed patient history This condition often first appears as acute pain in the hands and feet. Patients may also have: Blood tests Several blood tests can be used to look for SCD: Blood counts can reveal an abnormal Hb level in the range of 6 to 8 grams per deciliter. Blood films may show RBCs that appear as irregularly contracted cells. Sickle solubility tests look for the presence of Hb S. Hb electrophoresis Hb electrophoresis is always needed to confirm the diagnosis of sickle cell disease. It measures the different types of hemoglobin in the blood. How is sickle cell anemia treated? A number of different treatments are available for SCD: Rehydration with intravenous fluids helps red blood cells return to a normal state. Treating underlying or associated infections is an important part of managing the crisis, as the stress of an infection can result in a sickle cell crisis. An infection may also result as a complication of a crisis. Blood transfusions improve transport of oxygen and nutrients as needed. Packed red cells are removed from donated blood and given to patients. Supplemental oxygen is given through a mask. It makes breathing easier and

improves oxygen levels in the blood. Pain medication is used to relieve the pain during a sickle crisis. You may need over-the-counter drugs or strong prescription pain medication like morphine. Droxia, Hydrea helps to increase production of fetal hemoglobin. It may reduce the number of blood transfusions. Immunizations can help prevent infections. Patients tend to have lower immunity. Bone marrow transplant has been used to treat sickle cell anemia. Children younger than 16 years of age who have severe complications and have a matching donor are the best candidates. Home care There are things you can do at home to help your sickle cell symptoms: Use heating pads for pain relief. Take folic acid supplements, as recommended by your doctor. Eat an adequate amount of fruits, vegetables, and whole-wheat grains. Doing so can help your body make more RBCs. Drink more water to reduce the chances of sickle cell crises. Exercise regularly and reduce stress to reduce crises, too. Contact your doctor immediately if you think you have any type of infection. Early treatment of an infection may prevent a full-blown crisis. Support groups can also help you deal with this condition. What is the long-term outlook for sickle cell disease? The prognosis of the disease varies. Some patients have frequent and painful sickle cell crises. Others only rarely have attacks. Sickle cell anemia is an inherited disease. This can help you understand possible treatments, preventive measures, and reproductive options.

Chapter 3 : The Clinical Features of Sickle Cell Disease | JAMA Pediatrics | JAMA Network

Long term sickle cell anemia treatment 1. Stroke assessment: Doppler ultrasound on carotid artery to measure blood velocity and assess the risk of stroke in the brain calendrierdelascience.com high velocity, high risk --> regular transfusions.

They also are at higher risk for: Miscarriages Small-for-date or underweight babies Mental health As in other chronic diseases, people who have sickle cell disease may feel sad and frustrated at times. Sometimes they become depressed. People who have sickle cell disease may also have trouble coping with pain and fatigue, as well as with frequent medical visits and hospitalizations. Look for Treatment will discuss treatment-related complications or side effects. Living With will explain ways to manage complications of sickle cell disease. Diagnosis Your doctor may diagnose sickle cell disease based on the results from tests to confirm the results from various screening tests. This way, they can learn whether they carry a geneâ€"or have the traitâ€"for an abnormal hemoglobin that they could pass on to a child. Newborn screening When a child has sickle cell disease, early diagnosis is important to better prevent complications. The hemoglobin from this blood is then analyzed in special labs. If a baby is found to have sickle cell disease, health providers from a special follow-up newborn screening group contact the family directly to make sure that the parents know the results. The child is always retested to be sure that the diagnosis is correct. Newborn screening programs also find out whether the baby has an abnormal hemoglobin trait. If so, the parents are informed, and counseling is offered. These possibilities should be discussed with the primary care doctor, a blood specialist called a hematologist, or a genetic counselor. Prenatal screening Doctors can also diagnose sickle cell disease before a baby is born. Testing before birth can be done as early as eight to 10 weeks into the pregnancy. This testing looks for the sickle hemoglobin gene rather than the abnormal hemoglobin. Reminders Return to Signs, Symptoms, and Complications to review early signs and symptoms and complications of sickle cell disease. Return to Screening and Prevention to review how to screen for sickle cell disease. Treatment A blood and bone marrow transplant is currently the only cure for some patients who have sickle cell disease. After early diagnosis, the goal is health maintenance to prevent complications and medicines and treatments to manage complications, including chronic pain. Health maintenance to prevent complications Babies with sickle cell disease may see a hematologist, a doctor with special training in blood diseases such as sickle cell disease. For infants, the first sickle cell disease visit should take place before 8 weeks of age. If someone was born in a country that does not perform newborn screening, he or she might be diagnosed with sickle cell disease later in childhood. These people should also be referred as soon as possible for special care. Your doctor or medical team can help to prevent problems by taking certain steps: Educating families about the disease and what to watch out for Examining the person Performing tests Preventing infection In sickle cell disease, the spleen does not work properly or at all. This problem makes people who have sickle cell disease more likely to get severe infections. Penicillin In children who have sickle cell disease, taking penicillin two times a day has been shown to reduce the chance of having a severe infection caused by the pneumococcus bacteria. Infants need to take liquid penicillin. Older children can take tablets. Many doctors will stop prescribing penicillin after a child has reached the age of 5. All people who have had surgical removal of the spleen, called a splenectomy, or a past infection with pneumococcus should keep taking penicillin throughout life. Vaccines People who have sickle cell disease should receive all recommended childhood vaccines. They should also receive additional vaccines to prevent other infections. All people who have sickle cell disease should receive an influenza shot every year at the start of flu season. This vaccination should begin at 6 months of age. Only the inactivated vaccine, which comes as a shot, should be used in people who have sickle cell disease. The child should receive a booster vaccine three years after this series of shots, then every five years after that. This second vaccine is given after 24 months of age and again five years later. Adults who have sickle cell disease who have not received any pneumococcal vaccine should get a dose of the PCV13 vaccine. They should later receive the PPSV23 if they have not already received it or if it has been more than five years since they did. A person should follow these guidelines even if he or she is still taking penicillin. Screening tests and evaluations

Height, weight, blood pressure, and oxygen saturation Doctors will monitor height and weight to be sure that a child is growing properly and that a person who has sickle cell disease is maintaining a healthy weight. When a person who has sickle cell disease has high blood pressure, it needs to be treated promptly, because it can increase the risk of stroke. Oxygen saturation testing provides information about how much oxygen the blood is carrying. Blood and urine testing People who have sickle cell disease need to have frequent lab tests. Blood tests can tell your doctor whether you have another health problem, such as anemia or organ damage, so that it can be treated early. Urine testing can help to detect early kidney problems or infections. This study can find out whether a child is at higher risk for stroke. When the test results are abnormal, regular blood transfusions can decrease the chances of having a stroke. The child is awake during the TCD exam. The test does not hurt at all. The TCD machine uses sound waves to measure blood flow, like the ultrasound machine used to examine pregnant women. These exams can detect sickle cell disease-related problems of the eye. Regular exams can help doctors find and treat problems early to prevent loss of vision. A patient should see his or her doctor right away for any sudden change in vision. Pulmonary hypertension Doctors have different approaches to screening for pulmonary hypertension. This is because studies have not given clear information as to when and how a patient should receive the screening. People who have sickle cell disease and their caretakers should discuss with their doctors whether screening makes sense for them. Cognitive screening Patients who have sickle cell disease can develop cognitive problems that may be hard to notice early in life. Sometimes these problems are caused by silent strokes that can only be seen with magnetic resonance imaging MRI of the brain. People who have sickle cell disease should tell their doctors or nurses if they have cognitive problems, such as difficulties learning in school, making decisions, or organizing their thoughts. Patients can be referred for cognitive testing. This testing can identify areas in which a person could use extra help. Children who have sickle cell disease and who have cognitive problems may qualify for an Individualized Education Program, or IEP. An IEP is a plan that helps students reach their educational goals. Adults may be able to enroll in vocational rehabilitation programs that can help them with job training. Education and guidance Doctors and other providers will talk with people who have sickle cell disease and their caretakers about complications and also review information at every visit. Because there are many things to discuss, new topics are often introduced as a child or adult reaches an age when that subject is important to know about. Doctors and nurses know that there is a lot of information to learn, and they do not expect people to know everything after one discussion. People who have sickle cell disease and their families should not be afraid to ask questions. Topics that are usually covered include: They should try to feel for the spleen daily and more frequently when the child is ill. If they feel that the spleen is bigger than usual, they should call the care provider. How to recognize and manage pain The importance of regular medical visits, screening tests, and evaluations Transitioning care When children who have sickle cell disease become adolescents or young adults, they often need to transition from a pediatric care team to an adult care team. This period has been shown to be associated with increased hospital admissions and medical problems. There seem to be many reasons for this. Some of the increased risk is directly related to the disease. As people who have sickle cell disease get older, they often develop more organ damage and more disabilities. The shift in care usually occurs at the same time that adolescents are undergoing many changes in their emotional, social, and academic lives. The transition to more independent self-management may be difficult, and following treatment plans may become less likely. Compared with pediatrics, there are often fewer adult sickle cell disease programs available in a given region. This makes it more difficult for a person who has sickle cell disease to find appropriate doctors, particularly doctors with whom they feel comfortable. To improve use of regular medical care by people who have sickle cell disease and to reduce age-related complications, many sickle cell disease teams have developed special programs that the make transition easier. Such programs should involve the pediatric and adult care teams. The programs should also start early and continue over several years. Managing some complications of sickle cell disease Acute pain Each person who has sickle cell disease should have a home treatment regimen that is best suited to their needs. The providers on the team usually help a patient develop a written, tailored care plan. If possible, the person who has sickle cell disease should carry this plan with them when they go to the emergency room. When an acute crisis is just starting, most doctors will advise the patient to drink lots of

fluids and to take a non-steroidal anti-inflammatory NSAID pain medicine, such as ibuprofen. When a person has kidney problems, acetaminophen is often preferred. If pain persists, many patients find that they need a stronger medicine. Combining additional interventions, such as massage, relaxation methods, or a heating pad, may also help. Some patients may be able to return home once their pain is under better control. In this case, the doctor may prescribe additional pain medicines for a short course of therapy. Patients often need to be admitted to the hospital to fully control an acute pain crisis. When taken daily, hydroxyurea has been found to decrease the number and severity of pain episodes.

Chapter 4 : Haemophilus influenzae | Hib | Clinicial Features | CDC

These include sickle cell anemia (homozygous sickle mutation), sickle beta thalassemia, hemoglobin SC disease, and others. The clinical manifestations of SCD are protean. The major features are related to hemolytic anemia and vaso-occlusion, which can lead to acute and chronic pain and tissue ischemia or infarction.

Summary What is sickle cell disease? Sickle cell disease SCD is a group of inherited red blood cell disorders. If you have SCD, there is a problem with your hemoglobin. Hemoglobin is a protein in red blood cells that carries oxygen throughout the body. With SCD, the hemoglobin forms into stiff rods within the red blood cells. This changes the shape of the red blood cells. The cells are supposed to be disc-shaped, but this changes them into a crescent, or sickle, shape. The sickle-shaped cells are not flexible and cannot change shape easily. Many of them burst apart as they move through your blood vessels. The sickle cells usually only last 10 to 20 days, instead of the normal 90 to days. Your body may have trouble making enough new cells to replace the ones that you lost. Because of this, you may not have enough red blood cells. This is a condition called anemia , and it can make you feel tired. The sickle-shaped cells can also stick to vessel walls, causing a blockage that slows or stops the flow of blood. The lack of oxygen can cause attacks of sudden, severe pain, called pain crises. These attacks can occur without warning. If you get one, you might need to go to the hospital for treatment. What causes sickle cell disease? The cause of SCD is a defective gene, called a sickle cell gene. People with the disease are born with two sickle cell genes, one from each parent. People with sickle cell trait are generally healthy, but they can pass the defective gene on to their children. Who gets sickle cell disease? About 1 in 13 African American babies is born with sickle cell trait About 1 in every black children is born with sickle cell disease SCD also affects some people who come from Hispanic, southern European, Middle Eastern, or Asian Indian backgrounds. What are the symptoms of sickle cell disease? People with SCD start to have signs of the disease during the first year of life, usually around 5 months of age. Early symptoms of SCD may include Painful swelling of the hands and feet Fatigue or fussiness from anemia A yellowish color of the skin jaundice or the whites of the eyes icterus The effects of SCD vary from person to person and can change over time. Most of the signs and symptoms of SCD are related to complications of the disease. They may include severe pain, anemia, organ damage, and infections. How is sickle cell disease diagnosed? A blood test can show if you have SCD or sickle cell trait. All states now test newborns as part of their screening programs, so treatment can begin early. People who are thinking about having children can have the test to find out how likely it is that their children will have SCD. Doctors can also diagnose SCD before a baby is born. That test uses a sample of amniotic fluid the liquid in the sac surrounding the baby or tissue taken from the placenta the organ that brings oxygen and nutrients to the baby. What are the treatments for sickle cell disease? The only cure for SCD is bone marrow or stem cell transplantation. Because these transplants are risky and can have serious side effects, they are usually only used in children with severe SCD. For the transplant to work, the bone marrow must be a close match. Usually, the best donor is a brother or sister. There are treatments that can help relieve symptoms, lessen complications, and prolong life: Antibiotics to try to prevent infections in younger children Pain relievers for acute or chronic pain Hydroxyurea, a medicine that has been shown to reduce or prevent several SCD complications. It increases the amount of fetal hemoglobin in the blood. This medicine is not right for everyone; talk to your health care provider about whether you should take it. This medicine is not safe during pregnancy. Childhood immunizations to prevent infections Blood transfusions for severe anemia. If you have had some serious complications, such as a stroke , you may have transfusions to prevent more complications. There are other treatments for specific complications. To stay as healthy as possible, make sure that you get regular medical care, live a healthy lifestyle, and avoid situations that may set off a pain crisis.

Chapter 5 : Sickle Cell Disease | Sickle Cell Anemia | MedlinePlus

A 3-year comparative study of the clinical presentation of sickle cell disease in the high and low altitudes of the Assir Province of the Kingdom of Saudi Arabia is reported.

Sickle cell disease encompasses several entities: All of these manifestations of sickle cell disease can produce many of the clinical features of sickle cell anemia, but this paper will concentrate on sickle cell anemia SS. Sickle cell anemia is a genetic disorder, inherited in an autosomal recessive fashion. In sickle cell anemia, a single gene mutation results in the substitution of valine for glutamic acid in the sixth amino acid position of the beta globin chain, creating hemoglobin S. Under conditions of deoxygenation, hemoglobin S forms a poorly soluble tetramer that that causes red blood cells to deform into sickle shapes. Severe, chronic hemolytic disease, as well as various manifestations of vaso-occlusive disease, characterize sickle cell disease. Sickle cell disease can be diagnosed in patients of any age by hemoglobin electrophoresis revealing hemoglobin with mobility between Hb A and Hb A2 , isoelectric focusing, or DNA analysis. At present, all 50 states mandate newborn screening for the presence of sickle cell disease. Such a program has permitted early diagnosis of SCD, which â€" along with the implementation of a comprehensive system of care â€" has decreased the incidence of pneumococcal sepsis in young infants with SCD. The following manifestations are common in patients with SCD. Infection - due to RBC sickling, progressive infarction of the spleen is nearly universal in patients with SCD typically by age 4. Functional asplenia results, leaving patients susceptible to multiple bacterial infections, particularly encapsulated organisms such as S. Pneumococcal sepsis is the particular risk in these patients. Vaso-occlusive pain episodes â€" the most common clinical manifestations of sickle cell disease. The first manifestation is frequently dactylitis hand-foot syndrome , severe pain that occurs in the hands and feet, often in toddlers. Pain can also occur in other locations, such as the arms, legs, chest and ribs, and frequently lasts from two to seven days. Splenic sequestration â€" this is a common cause of morbidity and mortality in young children with sickle cell disease. The diagnosis is based on the sudden enlargement of the spleen accompanied by sharp drop in hematocrit and rise in the reticulocyte count. The enlargement of the spleen is due to entrapped blood. This can happen precipitously and circulatory compromise can occur. Acute chest syndrome â€" acute pulmonary injury is the leading cause of death in patients with sickle cell disease. Acute chest syndrome is defined as a new pulmonary infiltrate in combination with chest pain, fever or respiratory symptoms. Neurologic sequelae â€" neurologic complications such as stroke and hemorrhage can occur in children with SCD. Focal neurologic findings should alert the physician to possible neurologic issue. Anemia â€" patients with HbSS have a hemolytic anemia, which is generally well-tolerated unless a splenic sequestration episode or aplastic crisis occurs. These complications cause an acute drop in the hemoglobin level. Aplastic crisis is most often the result of infection with Parvovirus B19, which results in a temporary cessation of RBC production. Renal and Genitourinary â€" hematuria, renal failure, and priapism may occur. Avasuclar Necrosis â€" AVN of the femoral and humeral heads is a common manifestation of sickle cell disease, and is caused by multiple vaso-occlusive episodes in the bone. Cholelithiasis â€" due to severe hemolysis, patients with SCD can develop cholelithiasis and acute cholecystitis due to the formation of pigmented gallstones. A life-long cure for SCD is available only through hematopoietic stem cell transplantation, which carries its own toxicities and risks. Currently it is limited in use and reserved for patients under 16 years of age. Health Maintenance Much of the treatment of sickle cell disease is dependent on early diagnosis and continual preventative care. Children diagnosed with SCD should be referred to an early intervention program. The maintenance of full immunization status is essential, and additional vaccinations against pneumococcus and influenza virus are necessary. The valent conjugate vaccine series for pneumococcus should be started at 2 months, and the valent vaccine should be given at 2 years. The influenza vaccine should be given yearly. Prophylactic oral penicillin should be started at 4 months of age, dosed at mg BID until age 2; in children aged , oral penicillin should be dosed at mg BID. Folic acid supplementation should be started at 1 year of age. Hydroxyurea acts by increases production of HbF. In general, hydroxyurea is often used in children over two years of age who suffer from frequent vaso-occlusive pain events including

dactylitis , have a history of acute chest syndrome, or have severe symptomatic anemia. Caregivers of SCD patients should be counseled about signs and symptoms of illness. Additionally, parents should be instructed to seek immediate attention for the following signs: Broad spectrum antibiotics frequently ceftriaxone should be started while awaiting results of the cultures. Vaso-occulsive pain events â€" adequate pain control for the SCD patient is an important goal, but is challenging to achieve. In young children, or in patients with mild pain, Ibuprofen or Tylenol, with or without codeine, is preferred. Older children or children with more sever pain may require stronger narcotic analgesics. In the adolescent, a PCA pump patient controlled analgesia is often a good choice, as it allows the patient to self-titrate the medication. Splenic sequestration â€" severe sequestration can result in severe hypovolemia and circulatory compromise. An emergent red cell transfusion and fluid recussitation can be life saving measures. Acute-chest syndrome â€" the child with acute chest syndrome should be hospitalized and monitored intensely, often in an ICU. The patient should be evaluated with serial CXRs and should be treated with oxygen or exchange transfusions as necessary. The patient should be started on a cephalosporin and a macrolide to cover infectious causes of acute chest syndrome. Stroke â€" strokes in children with SCD often occur without warning, but can be preceded by a severe headache or focal neurologic deficit. The only treatment currently available for strokes is the use of monthly exchange transfusions to decrease the amount of HbS. Anemia â€"is usually well tolerated, except in the event of splenic sequestration or aplastic crisis. In these cases, red cell transfusion is indicated.

Chapter 6 : Sickle Cell Disease | Pediatrics Clerkship | The University of Chicago

Pain crises constitute the most distinguishing clinical feature of sickle cell disease and are the leading cause of emergency department visits and hospitalizations for affected patients. Approximately half the individuals with homozygous Hb S disease experience vaso-occlusive crisis.

Treatment involves a number of measures. L-glutamine use was supported by the FDA starting at the age of 5 as it decreases complications. It has therefore been recommended that people with sickle cell disease living in malarial countries should receive lifelong medication for prevention. However, the frequency, severity, and duration of these crises vary tremendously. Painful crises are treated symptomatically with pain medications ; pain management requires opioid administration at regular intervals until the crisis has settled. For more severe crises, most patients require inpatient management for intravenous opioids; patient-controlled analgesia PCA devices are commonly used in this setting. Diphenhydramine is also an effective agent that doctors frequently prescribe to help control itching associated with the use of opioids. Should the pulmonary infiltrate worsen or the oxygen requirements increase, simple blood transfusion or exchange transfusion is indicated. The patient with suspected acute chest syndrome should be admitted to the hospital with worsening A-a gradient an indication for ICU admission. Hydroxyurea had previously been used as a chemotherapy agent, and there is some concern that long-term use may be harmful, but this risk has been shown to be either absent or very small and it is likely that the benefits outweigh the risks. Bone marrow transplants are the only known cure for SCD. Ideally, a close relative allogeneic would donate the bone marrow necessary for transplantation. Avascular necrosis[edit] When treating avascular necrosis of the bone in people with sickle cell disease, the aim of treatment is to reduce or stop the pain and maintain joint mobility. Increased risk of severe bacterial infections due to loss of functioning spleen tissue and comparable to the risk of infections after having the spleen removed surgically. These infections are typically caused by encapsulated organisms such as Streptococcus pneumoniae and Haemophilus influenzae. Daily penicillin prophylaxis is the most commonly used treatment during childhood, with some haematologists continuing treatment indefinitely. Patients benefit today from routine vaccination for S. Cerebral infarction occurs in children and cerebral haemorrhage in adults. Silent stroke is probably five times as common as symptomatic stroke. Avascular necrosis aseptic bone necrosis of the hip and other major joints may occur as a result of ischaemia. During pregnancy, intrauterine growth retardation , spontaneous abortion , and pre-eclampsia Chronic pain: Even in the absence of acute vaso-occlusive pain, many patients have unreported chronic pain. Where malaria is common, carrying a single sickle cell allele trait confers a heterozygote advantage: The parents each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. This happened in predominant areas of malarial cases. As of all 50 states include screening for sickle cell disease as part of their newborn screen. Since , neonatal screening of SCD has been performed at national level for all newborns defined as being "at risk" for SCD based on ethnic origin defined as those born to parents originating from sub-Saharan Africa, North Africa, the Mediterranean area South Italy, Greece and Turkey , the Arabic peninsula, the French overseas islands and the Indian subcontinent. As the number of carriers is only estimated, all newborn babies in the UK receive a routine blood test to screen for the condition. In , a law was passed requiring couples planning to get married to undergo free premarital counseling. These programs were accompanied by public education campaigns. Irons â€" , intern to the Chicago cardiologist and professor of medicine James B. Herrick â€" , in Irons saw "peculiar elongated and sickle-shaped" cells in the blood of a man named Walter Clement Noel, a year-old first-year dental student from Grenada. Noel had been admitted to the Chicago Presbyterian Hospital in December suffering from anaemia. He died of pneumonia in and is buried in the Catholic cemetery at Sauteurs in the north of Grenada. In , the introduction of haemoglobin electrophoresis allowed the discovery of particular subtypes, such as HbSC disease. Social Security[edit] Effective September 15, , the U. Social Security Administration issued a Policy Interpretation Ruling providing background information on sickle cell disease and a description of how Social Security evaluates the disease during its adjudication process for disability claims. In humans, using hydroxyurea to stimulate the production

of HbF has been known to temporarily alleviate sickle cell disease symptoms. The researchers demonstrated that this gene therapy method is a more permanent way to increase therapeutic HbF production. The clinical trials will assess the safety and initial evidence for efficacy of an autologous transplant of lentiviral vector-modified bone marrow for adults with severe sickle cell disease. Current nomenclature calls for counting the methionine as the first amino acid, resulting in the glutamic acid residue falling at position 7. Many references still refer to position 6 and both should likely be referenced for clarity.

Chapter 7 : The Clinical Features of Sickle Cell Disease: Clinical Studies | JAMA Ophthalmology | JAMA N

The pathophysiology and clinical features of sickle cell disease at sea level have been extensively studied and reported from different parts ofthe world[],including Saudi Arabia [].

Chapter 8 : Sickle cell anemia - Symptoms and causes - Mayo Clinic

The molecular biochemistry, the physiochemical basis of sickling, tests for sickle hemoglobin, demography, genetics, and history of sickle cell disease are concisely, but authoriatively, presented.

Chapter 9 : Pathobiology Clinical Features, And Management Of Sickle Cell Disease | Clinical Gate

Sickle-cell disease is an inherited blood condition common among, but not confined to, peoples of Equatorial African ancestry. The gene for sickle haemoglobin (HbS) results in the substitution of valine for the glutamic acid normally present at the sixth position from the amino terminus of the Î² chain of haemoglobin.