

BLOOD DISORDER DISEASES

INTRODUCTORY

Just as characteristics like hair or eye colour are passed on in families through the genes, in the same way certain abnormalities can be genetically inherited. If either you or your partner have some sort of inheritable condition such as sickle cell disease, abnormalities of the blood type and Rhesus factors. If either of you have a relative with such as condition then you may want to know what sort of risk, if any, you would be taking by having a baby. It is important to understand this risk as far as possible before you marry or conceive.

SICKLE CELL DISEASE

Sickle cell disease is a chronic, severe hereditary disease. It is referred to as rheumatism in Ghana therefore the various ethnic groups in Ghana call it by different names such as “Chweahwechwe “ in Ga, “Nweewee” in Fante, “Ahotutuo” in Ashanti, “Chichi” in Ewe, “Amosani in Hausa. Normally there are three types of haemoglobin A, A2 and F. The F is what we have at the foetal stage.

Sickle cell disease occurs in two forms:

- a) The sickle cell trait i.e. carriers who usually do not manifest the symptoms of sickling but there is 50% chance that their offspring's will inherit the sickle cell anaemia.
- b) It results when the individual inherits one normal haemoglobin gene: from one parent and abnormal gene (s) from another parent. Thus he has haemoglobin AS.

c) Sickle cell Anaemia is a sickle cell disease in which both genes are abnormal. The common types are the following: SS, SC, SF and S Thalassaemia.

Description of the sickle cell:

The sickle cell disease makes the red blood cells to assume a crescent or sickle or half moon shape instead of the normal round shape. The sickle red cells cannot pick sufficient oxygen from the lungs. This explains why they always feel tired. The life span of the sickle red blood cell is reduced from the normal 120 days to 20 days as they are easily destroyed. The sickle cells increase when there is oxygen tension in the blood. This is the reason why sicklers are advised to avoid trips to higher altitudes.

Incidence:

The disease affects both male and females and occurs exclusively in Africans, (Negroid race) West Indians and less often, Asian people. The other blood disease S Thalassaemia can affect people from Middle East, Mediterranean and India also. It is known that 1-5 of all West African will inherit abnormal haemoglobin.

Clinical Manifestation:

ON THE CHILD

- 1) Clinical symptoms appear around the end of the first year of life, this signifies the period by which the foetal haemoglobin is replaced.
- 2) The main features of the disease in infancy are swelling and pain of hands, fingers, feet and toes, (hand-foot syndrome) paleness and rarely jaundice (yellowish discolouration of the eyes)

- 3) From the second year of life, the disease is characterised by repeated attacks of fever, jaundice, anaemia, pain in the limbs and abdomen due to infarcts (foreign bodies) in the bones and mesenteric capillaries (blood vessels of the intestines). There is enlargement of the spleen. In long standing cases, the heart is dilated and has murmurs (noises)
- 4) Teenage sicklers may have chronic ulcers especially above the medial Malleous. Healing is very slow and last for 4 months.
- 5) There is constant destruction of the red blood cells leading to chronic anaemia. The Hb level in sickle cell anaemic, ranges from 6-8g/dl. The person then complains of palpitation, tiredness, and dizziness and looks pale.
- 6) The constant destruction of the red blood cells leads to accumulation of bilirubin in the blood resulting in jaundice. This is common with the SS patients.
- 7) There is enlargement of liver and spleen due to overworking and pooling of blood from these organs. These may be found in SS, Sc and S Thalassaemia.
- 8) Sickle-cell disease patients are prone to infections of the bones known as osteomyelitis. Again they are prone to infections caused by Gram-negative organisms eg typhoid. Others include; upper respiratory tract infections and urinary tract infections. The SS patients are prone to malaria whiles it is believed that AS traits are resistant to malaria
- 9) There is growth disturbance (stunted or very tall).
- 10) Some also experience unexplained blood in urine due to problems of the kidneys.
- 11) In men, there may be painful erection of the male organ (priapism).
- 12) In women-irregular periods are common.

- 13) Again there may be bleeding from the nose (epistaxis).
- 14) Hip trouble-Aseptic necrosis (the head of the thigh bone may cause chronic pain and difficulty in walking and may require attention of orthopaedic specialist).
- 15) Eyes may be affected leading to blindness

Effects of sickle cell disease on Pregnancy

- This carries a high mortality (both mother and baby should be treated with care).
- Severe Anaemia should be treated vigorously.
- Increase incidence of spontaneous abortions, intrauterine growth retardation and intrauterine foetal deaths.
- Post Partum Haemorrhage.

Management:

- 1) Pregnant sicklers must attend antenatal clinic regularly under the supervision of a specialist. Hospital delivery should be ensured.
- 2) The nutrition should be good.
- 3) Anti malaria prophylaxis should be enforced to prevent malaria.
- 4) Fatigue or tiredness must be avoided.

Diagnosis:

- 1) If there is hand-foot syndrome in a child of 4 years it is suggestive.
- 2) Family History.
- 3) Blood examination for sickling. If positive, etc electrophoresis may be done to determine the type of sickle cell disease.

Sickle Cell Crisis

A patient with sickle cell disease may get into a critical situation known as crisis. Here there is severe pain, jaundice, there is excessive destruction of the red blood cells leading to thickening of the blood which may result in overworking of the heart, or there may be dislodgement of the clots leading to death.

Things that may trigger Crisis

- 1) Fever or malaria
- 2) Infections-sore throat, coughs, diarrhoea and urinary tract infection.
- 3) Vaccination
- 4) Injury
- 5) Pregnancies or deliveries
- 6) Blood Transfusion
- 7) Exposure to high altitude
- 8) Alcoholic intoxication
- 9) Exposure to cold

A) Preventive measures

- Counselling of young people well before they begin to consider marriage.
- Young people in our schools and work places must be taught that sickle cell disease results from two people who carry the sickle cell gene having children together.
- It is therefore important for each person to know his/her haemoglobin genotype. Those with the sickle cell gene must make

it a point not to marry similar people. This counselling must be reinforced at pre-marital counselling.

- There must be regular education through the media and in all established communal gathering for people to know their haemoglobin genotypes.
- Sickle cell disease patients, especially the female ones must make it a point to attend regular medical check-up so that they are in the best state of health prior to getting pregnant. This reduces the risks of maternal death.
- Sickle cell disease pregnant women must report early for antenatal care. They must start being seen by a doctor as soon as they miss their period.
- Nourishing diet and fluids must be encouraged.
- Daily folic acid supplements
- Regular anti malarial drugs with prompt treatment of infection.

In conclusion, sickle cell disease cannot be treated but it can be controlled for individual to enjoy life.