1. What is sickle cell disease?

Sickle Cell is a genetic disorder affecting the red blood cells of blood which affects every system or part of the body. But it is well-known for the typical, excruciating and crushing bone and joint paints earning it its local name of "*ahotutuo*" in Twi.

2. What are red blood cells and haemoglobin?

Red blood cells, RBCs are usually 'born' from our bone marrows with a flexible, doughnut shape containing a *red* pigment (hence, the name *red* blood cells) called *haemoglobin*. These properties of the RBCs and the haemoglobin enable them to perform their crucial function of carrying oxygen from the lungs to the rest of the body. Oxygen is part of the air we breathe. It is fuel that powers our bodily functions.

3. Do red blood cells live forever once they are 'born'?

Red blood cells usually live for 3 months in our blood. Our bone marrows make new ones to replace the dead ones which are removed by an internal organ called the spleen, usually hidden under our ribs on the left side of our bodies

4. How does sickle cell disease affect our red blood cells?

Sickle cell disease unfortunately, distorts the shape, flexibility and the red pigment of the RBCs. Sickle cell disease makes the RBCs stiff or rigid, sticky and to assume the shape of a banana or a crescent (quarter moon). Sickle is a crescent-shaped farming implement which was used to harvest rice in the olden days.

5. What causes sickle cell pain?

Under certain conditions (called triggers) the abnormal stiff, sickled RBCs clog or stick together and block the pipes (called blood vessels) which carry blood around the body. Scientists call this process *sickling*. This results in poor oxygen supply to the affected body part(s). Sickling can happen in any part of the body.

When it happens in the bones it causes very severe pain; in the lungs it causes severe shortness of breath and chest pain; in the brain it can lead to strokes; in males it can cause an abnormally, sustained painful erection; etc.

6. What are the common triggers?

These include dehydration (not drinking enough water and/or sweating too much); cold weather; strenuous exercises; fever (high body temperature); infections like malaria; and even pregnancy

7. What are the consequences of having abnormally sickled cells?

The abnormal sickled red blood cells, unlike their normal counterparts, survive for only 2 to 3 weeks. Their breakdown (deaths) turns urine dark; cause low hemoglobin levels seen as paleness and tiredness; yellow staining of the eyes (jaundice); and the abnormal enlargement of spleen (body organ that cleanses the blood and helps fight infections) seen as a bloated tummy.

8. Is sickle cell type the same as my blood group?

Blood group is different from sickle cell type. Your blood group (named O, A, B or AB) is named after some proteins on the surface of your red blood cells. Sickle cell type (A, S, C, etc.) on the other hand refers to the gene (*supervisor*) responsible for making the haemoglobin in RBCs. Blood groups and sickle cell types are completely different.

9. What do the letters "A", "S", "C" mean?

These are 'names' scientists have given to the various kinds of haemoglobins in red blood cells. These haemoglobins are the products or results or creations of genes (supervisors) that control them. The normal gene is produces hemoglobin "A". In sickle cell there are specific 'mistakes' in these genes leading to specific types of abnormal haemoglobins such as haemoglobins "S" and "C".

In Ghana, among these abnormal haemoglobins, the "S" type is the commonest by far followed by the "C" type. There are several other abnormal haemoglobin types apart from these two.

10. What does it mean to be "SS" or "AS"?

Each human being must have two (a pair of) daughters from the gene Each parent (father and mother) contributes one half of the gene in making a child. If you have one "A" (for example "AS" or "AC") you are described as a *carrier* of the abnormal sickle cell gene; also known as *sickle cell trait*. In that case you do not show any signs of the disease.

But if none of the 'daughters' is "A" (for example "SS" or "SC" or "CC") then both haemoglobins are abnormal and you are described as having sickle cell disease. Those who specifically have the "SS" are described as *sickle cell anaemia*. Scientists refer to these various sickle cell types as *phenotypes* or *genotypes*. Generally, we refer to them as your sickle cell *status*.

11. Why should I know my sickle cell status?

Each parent passes one half of the pair of '*letters*' to their child. Knowing your sickle cell status is important so that you and your partner will know if the two of you are likely to pass on either abnormal ("S" or "C") or normal ("A") genes to your child. This is the only way to prevent sickle cell disease.

12. "My partner and I are both AS, can we marry"?

It is important to clearly distinction between ***marriage*** and producing children or ***reproduction***. Admittedly, marriages are expected to produce children. But the decision to marry is a mutual agreement between two competent and consenting adults.

Carriers of the abnormal sickle cell gene ('AS' or 'AC') who intend to produce children (whether as married couples or not) must however, be ***well informed*** about the genetic implications on their potential offspring or children as well as any remedies available to support them. Armed with this information such a couple can go on to make an ***informed decision***.

If two carriers, both 'AS' decided to have a child naturally, there is a 25% (1 in 4) chance that ***each pregnancy*** between such a couple, will result in a baby who is 'SS'. **Please note** that the '1 in 4' chance here, **does not** mean that assuming you decided to have 4 children, only one will be 'SS'; or that if you had one child who is 'SS' then all other children will not be 'SS'.

Science cannot as yet predict which pregnancy will result in an 'SS' outcome. That is simply left to chance. Also, be advised that we have very good clinical services and care for all persons including children with sickle cell disease including those with the 'SS' genotype.

12.1. So, can all our children be 'SS'?

Precisely. Similarly, if you decided to have more than one child with the same partner naturally, all of the children could be 'AA' or 'AS' or a mix of all three different genotypes. It is simply a matter of chance.

12.2. Can a pregnant carrier ('AS' mother) take any medicine to prevent transmission of the abnormal 'S' gene to the unborn baby?

Currently, there are no medical interventions or medicines available to prevent this. However, it is possible to safely determine the genotype of the unborn baby (while in the mother's womb). This is a highly specialized test requiring highly skilled expertise in a hospital setting. Once this determination is made, experts (genetic counselors) counsel parents as to the available options.

13. Is there a cure for sickle cell disease?

There are two treatments that can provide a cure for sickle cell diseases: 1. Bone Marrow Transplant and 2. Gene Therapy. These treatments are not without risks though, and their costs make them prohibitive and practically inaccessible to many people with sickle cell disease in Ghana. The next best thing to a cure for people with 'SS', if it works, is hydroxyurea therapy. Hydroxyurea therapy is well described in another publication.

14. How do I know if my child is 'SS'?

We recommend for all persons to know their sickle cell statuses to prevent the potential of producing children with 'SS'. We also strongly recommend for ***all children born in Ghana*** to undergo a routine newborn sickle cell test to determine their sickle cell statuses to ensure early diagnosis and further appropriate care for those with the disease.

Children with 'SS' who are not diagnosed early through newborn screening, only begin to show signs of the disease from about 3 to 4 months of age including painful, swollen hands and feet; infections (fevers); and at an older age, pale appearance; yellow staining or discoloration of eyes; bone pain; and passage of dark-colored urine. Thus, we recommend routine sickle cell screening test in our hospitals for all children whose sickle cell statuses are not known. Sickle cell disease is a recognized cause of death of children under 5 years who do not receive appropriate care.