

Communicating basic genetics to patients with sickle-cell disease

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SUMMARY This report describes an approach to explaining the basic genetics of sickle-cell disease to patients and their families in an uncomplicated yet sufficiently informative way. The areas covered include: the cause of the disease, its pathophysiology, and the importance of genetic counselling.

Introduction

The sickle-cell gene is known to be widespread throughout the world, occurring most frequently among black Africans. Confusion, ignorance and misconceptions about sickle-cell disease (SCD) are still common, particularly with regard to its cause and pathophysiology. Local belief and fanciful folklore add to the problem [1-5].

The cumulative effect of psychosocial stress may have a devastating effect on the families involved, and adversely affect the entire community in which they live. Families known to include many members with SCD may be stigmatized as having a curse on them [6, 7].

In a busy clinic or ward round, health carers rarely have enough time to explain the implications of the disease and how the patient should be cared for. Thus, many parents go home without a clear idea of what SCD is, or what it entails. Worse still, many doctors working in Saudi Arabia and neighbouring countries never (or very rarely) see cases of SCD, so when a patient

does present with the disease, they are not really in a position to give appropriate care to the affected patient, or guidance to the parents of the patient.

It is important to explain the basic facts about SCD to both professionals and lay people in simple language that avoids causing confusion or doubt. The educational aspects of genetic counselling involve breaking down complicated information about SCD into its basic essentials, without obscuring the necessary facts, and keeping it sufficiently interesting for those being counselled to retain the information [8].

This paper outlines an approach to explaining the basic genetics of SCD to patients.

Communicating with patients

Cause of the disease (genetics)

Two illustrations are used as visual aids (Figures 1 and 2). It is explained to the patient and parents that everyone has blood in their body, which is very important for the body, similar to the importance of petrol to

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a vehicle. When our blood is examined under a microscope, some people's blood looks different from others. Most people have blood that contains small objects shaped like a full moon (point to "A" in Figure 1). Such people are called AA. Some people have blood that contains new moon shapes (point to "S" in Figure 1). Such people are called "SS" or "sicklers". Some people have both full moons and new moons. They are described as "AS". Whether we have full moon shapes or new moon shapes, or both, depends on the shapes each of our parents has. We cannot change these shapes in our blood, although we can learn how to avoid having "SS" children ourselves.

Every child takes something from both the father and the mother. An SS child will take a new moon from his father and a new moon from his mother. This means that each of the child's parents must have at least one S in their blood; they are either AS or SS. At this stage show Figure 2, illustrating an SS child and parents [4,5,8].

Associated features (pathophysiology)

Anaemia

"Full moon" blood survives up to 120 days, whereas "new moon" blood survives just half of that period. Full moon blood can move freely in the body, whereas new moon cannot. It gets clogged up. New moon blood is not as strong as full moon blood. Therefore, a person with only new moons in their blood (SS) is often short of blood. (The point must also be made that people who are AS have no problems and are normal.)

The parents are shown the parts of the body in which to look for pallor, and we emphasize the need to take daily medicine (folic acid), which helps to build up the patient's blood.

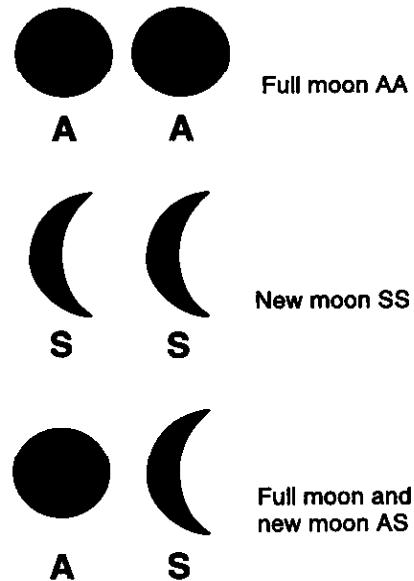


Figure 1 Diagram to illustrate the difference between the blood of people with sickle-cell disease and those without the disease

Illness

Environmental factors, including malaria, other infections, infestations and malnutrition can modify the manifestation of SCD. Illnesses such as malaria may break down the blood, leading to greater anaemia and yellow eyes (jaundice). Sometimes the child may need a blood transfusion. The control of malaria is a major factor in improving the morbidity and mortality of patients with SCD. Taking antimalarial precautions regularly will help to reduce the number of illnesses.

Painful crises: extreme individual variation

Some patients may have a crisis only once every few years, while others may have one per month, or even weekly. Painful crises are precipitated by factors such as infection, dehydration or deoxygenation (e.g. al-

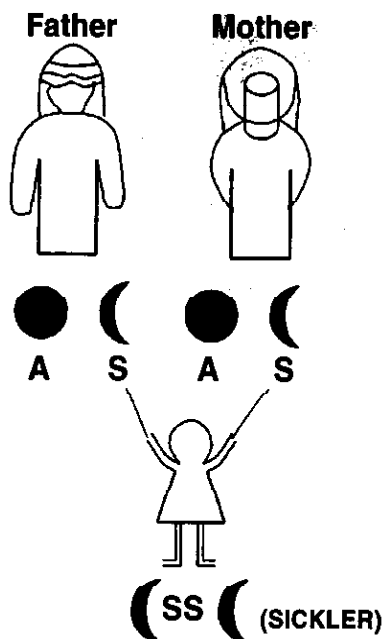


Figure 2 Diagram to illustrate the inheritance of sickle-cell disease

titude, operation, obstetrics delivery, exposure to cold and violent exercise).

Painful crises are caused by the new moon cells sticking together and causing a "road block" in the body. This can happen in any part of the body, often in the bones. Making sure the child drinks plenty of fluid will help to "unstick" S cells and remove the road block. A child who cannot drink enough must have a drip in the hospital [3]. The clinical course of sickle-cell anaemia is extremely variable, and is characterized by chronic haemolytic anaemia and various types of crises (e.g. abdominal, aplastic, splenic sequestration), which ultimately may lead to organ damage. The organs most frequently involved are the spleen, central nervous system, liver, kidney and heart.

Genetic counselling

A child is a product of a male and female partner. It must be gently emphasized that a child who is SS has inherited new moons from both parents, a fact, unfortunately, that many fathers quickly dismiss. An empathetic approach needs to be taken when communicating this idea to parents. Using the diagram, a couple can be led to understand that some of their children may be SS, and others AS or AA. Participants should be encouraged to raise issues as they wish.

In this part of the world, where marriages are arranged by parents, it is absolutely important to point out that an SS child should try to marry someone who is AA, in order to avoid having children who may be sicklers. Couples at risk of producing offspring susceptible to SCD may wish to consider using prenatal diagnosis and selective termination of affected pregnancies in order to ensure a healthy family. However, this is controversial because of the religious/ethical issues involved. It is important to avoid confusion in counselling, or to be seen to be promoting controversy in so sensitive an area as the religious aspects of health [4,8].

Other relevant advice

Patients should be advised to avoid "trigger" factors such as cold or violent exercise, and keep warm. They should regularly attend the sickle-cell clinic, so that progress can be monitored and problems detected early.

Frequently asked questions

Because of ignorance about SCD, parents and patients always have a lot to ask. Care must be taken when answering questions so as not to create fear. One must be tactful when responding to questions asked. Frequently asked questions, such as "Can this

be cured?"; "Can the blood be washed and replaced?"; "Does he/she need any special food?"; and "Is she/he going to die?", require tactful, sensitive responses. You must be frank and objective in answering but at the same time and you should try to be hopeful, realistic and optimistic. While acknowledging that many of the children are often sick, and some do die, it should also be stressed that survival to adulthood is possible with good medical attention, control of the feature(s) that trigger crises and good nutrition.

If the question of divorce is raised, you must avoid saying yes or no. Rather, reference should be made to the genetic counselling already given. The breaking up of the family is not the solution. Family support is extremely important to the SCD-affected child. The child's well-being and survival are optimized within a united family environment [8-10].

Discussion

The dissemination of information is an important factor in augmenting awareness programmes among professionals as well as lay people. It is very important for health workers not only to know the facts about an illness, but also to communicate these facts in a very simple way. This will help people improve their family health and focus attention on the main issues [8,11].

The inability of many health workers in our area to effectively communicate the facts about the S gene in the local language is a handicap to increasing awareness about the problem. One way to overcome this is to use concepts and symbols which the local population clearly relate to. People in this area are familiar with the moon, and can easily understand the symbols used in the approach suggested here. The moon is

of significance to Muslims, so the people can relate to and more easily understand and accept the discussion.

Once the facts about SCD are made clear, it takes less than a quarter of an hour to get the message across, even when an interpreter is involved. During the sickle-cell clinic day, a knowledgeable patient/parent from the community is always used to address the group, using the previously mentioned visual aids. The doctor should be close at hand to answer any questions from the gathering. This approach has worked extremely well in our area [3,12].

Effective communication is a two-way affair, involving listening just as much as talking. Group discussion enables people with different opinions to express their views, anxieties, and also to ask questions. This has proved to be particularly informative and stimulating as patients and parents share knowledge of how to care for themselves. However, opportunities for more individualized, one-to-one attention that some families may need for in-depth discussion of specific family situations, may be limited [8,9,13]. It is the duty of all health workers to be friendly and receptive, and to take an interest not only in the patient in the clinic, but the entire family as well. Inquiring about the well-being of the family creates a better atmosphere between the health worker and the child's family [10,12].

It is extremely important to make sure that all basic facts discussed during the session are clearly understood, and there is absolutely no doubt in the minds of the patient and parents about what has been discussed. The mode of inheritance must be re-emphasized, because a basic understanding of the genetic factors may affect many aspects of the patient's life. Failure to understand that the problem is inherited from both parents often leads to unfair accusation, victimiza-

tion and a sense of guilt of one spouse by another. It is the marriage that often suffers [4,6,7,8].

In this region, where marriages are arranged by parents, the desirability of selecting an AA partner should be stressed and seen to be understood. Couples who are carriers at risk of having affected children should be informed of their one-in-four chance of having an affected child in each pregnancy [5,11].

Only the S gene is mentioned in this paper. Variants such as C and D are not dealt with here. Other factors which can affect prognosis, for example Hb F level, red blood cell deformation, glucose-6-phos-

phate dehydrogenase haplotype and interaction with thalassaemia are also omitted, as is the gene of SB thalassaemia.

It was realized very early on that a haemoglobinopathy control programme should be an integral strategy combining optimum management of existing patients, community education in prevention, prospective heterozygote diagnosis, genetic counselling and prenatal diagnosis. Efforts need to be made to ensure that such a control programme presents its message in a way that is easily understood and relevant to this region, and that the members of the target population are in no doubt as to the facts.

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