HEALTH & Beauty
New Vision YOUR WELLNESS GUIDE
Monday, April 16, 2018 27

HAEMOPHILIA THE MALE-ONLY DISEASE

By Vivian Agaba

Uganda will joins the rest of the World tomorrow to commemorate World Haemophilia Day. However, experts say little is still known about the disease.

This year, the day is being commemorated under the theme: *Sharing knowledge.*

Dr Philip Kasirye, a paediatrician at Mulago, defines haemophilia as an incurable, hereditary (genetic) bleeding disorder in which the affected person's blood does not clot normally because it lacks sufficient blood-clotting proteins.

This means that the blood cannot clot or form a mass at the site of a wound or injury. The disorder occurs because certain blood clotting factors are missing or do not work properly. Because a clot does not form, even a simple cut can cause extensive bleeding.

Haemophilia is caused by a gene mutation, which is a change in the genetic material on the X-chromosome, a thread-like structure inside human cells that contains genetic information that is passed down through families.

Kasirye says there are two main types of haemophilia – haemophilia A, which is as a result of Factor VIII deficiency. Majority of patients fall under category A (75-80%). There is also Haemophilia B, which is due to factor IX (9) deficiency with a 10-15% of patients.

"The hemophilic individual either totally lacks or have reduced amount of factors, especially 8 and 9, which are the commonest. This means they do not adequately form fibres, and, therefore, cannot form a strong clot to control bleeding," he explains.

Generally, the disease is known to present in males. This is because the genes for the disease are carried on an X chromosome, given the Y chromosome in men is dominant. Females have two copies of the X chromosomes, while males have one X and one Y chromosomes. For girls or females, the extra X chromosome in the mother is protective if it does not carry the haemophilia gene, hence they are largely carriers. This means carriers pass the gene onto their male children.

However, it is also important to note that sometimes, haemophilia happens spontaneously and without a carrier mother. Some genetic changes may occur and a child is born with haemophilia yet none of the members from the lineage has the condition, Kasirye explains.

Prevalence

Though no research has been done in the country about the disease, Agnes Mavis Kisakye, the executive director of the Haemophilia Foundation of Uganda, says out of 10,000 children in Uganda, 1% have the disease, and this

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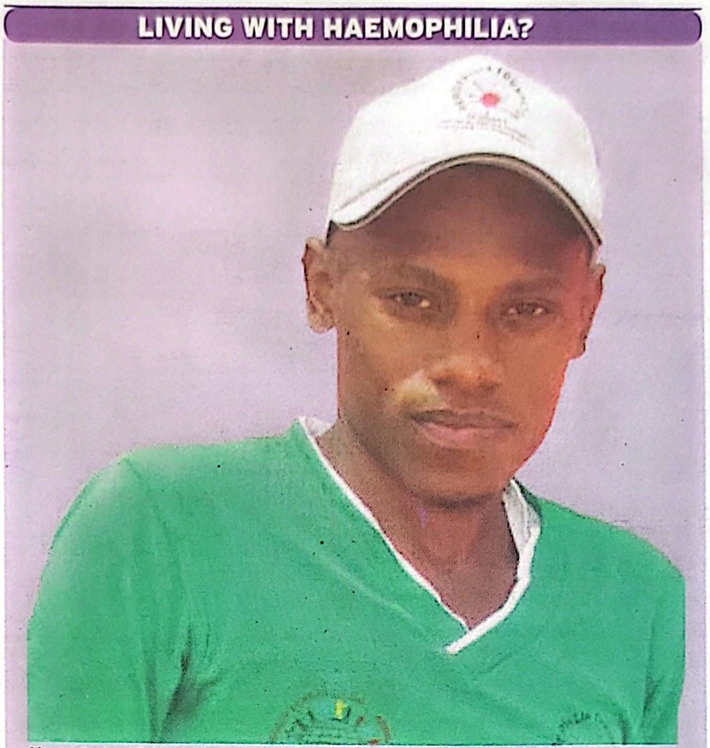
LIVING WITH HAEMOPHILIA?



Nangulu has joined the awareness team in Uganda. Photo by Vivian Agaba

It was in 2016, when Peter Nangulu, 23, a student at YMCA in Kampala found out that he had haemophilia, a condition that leads to uncontrollable bleeding. This was after he had undergone tests at a clinic in Entebbe.

Though he had all the signs of the disease from childhood, his parents could not figure out what the problem was. Just like any other unfamiliar health conditions, Nangulu says his parents and community members thought he was bewitched.

Out of six children; four boys and two girls, Nangulu is the only one with the disease.

It was at the age of four that the disease began to manifest. Nangulu who hails from Butaleja district was taken to various health facilities where he was wrongly diagnosed with sickle cell disease. "I started experiencing body swellings, especially in the joints and legs after walking long distances. Whenever I played with my peers and fell down and sustained minor injuries, I would bleed for days," he recalls.

"This disease put many restrictions on my life," he adds.

Nangulu says there is a lot of stigma that comes with the disease. He recalls one of the doctors used to call him 'delicate', while other people thought he would die young.

However, ever since he was diagnosed and started treatment at Mulago Hospital, the episodes of bleeding have somewhat stopped.

He goes to the facility once a week to have a factor put into his blood through an injection.

Symptoms of haemophelia

The signs of haemophilia include prolonged bleeding, especially in joints and muscles, big bruises. It can also manifest through sudden bleeding inside the body for no clear reason, bleeding for a long time after getting a cut such as removing a tooth, or after surgery such as circumcision or after an accident.

Normally, after a person has sustained an injury, it is supposed to take about eight to 15 minutes for a clot to form on the affected area, thus stopping the bleeding. However, in individuals with haemophilia, the bleeding may take hours, a day or even days.

Dr Henry Ddungu, a consultant hematologist at the Uganda Cancer Institute in Mulago, explains that if an individual with haemophilia is playing and falls, they will develop bruises, which are usually reddish or purple and will start bleeding under the skin.

"They can also present with general body weakness and headaches. The person may also pass urine with blood, and in severe cases, the person may also vomit blood, especially if the bleeding is in the brain," Ddungu asserts.

Though the condition is rare among females, a woman who has the condition can bleed heavily and for more than a week during the menstrual periods. They should, therefore, go to the health facilities for testing.

Dr Philip Kasirye, a paediatrician at Mulago, adds that if a child bleeds internally, especially in the vital organs such as the brain, lungs, abdomen, kidneys, it can be life-threatening or lead to death.

"If a boy complains of severe abdominal pain, severe headaches has convulsions or fits, the child has difficulties in breathing, severe vomiting, rush him to a hospital. This is because it could be that the child is bleeding internally in the brain, lungs or abdomen," Kasirye advises. Parents that should suspect their sons have haemophilia if there is a family history of prolonged bleeding among male relatives with deformed knees.

HEALTH VIEWPOINT

Caution on over bleeding in men

When we hear the term "over bleeding", many of us will think of a female. However, there is one known condition that can make males over bleed and it is life-threatening. This condition is known as haemophilia, whose day is commemorated every April 17. As we get ready to mark the day tomorrow, I encourage you to read our big story to find out what signs you should not overlook in a male and what to do to ensure the person enjoys considerable good health.

In addition, I encourage you to continue communicating us and let us know the issues you want us to sensitise the readers about.

I wish you good health!

Lillian Namusoke Magezi

Activities

There will be several activities in some parts of the country to mark the haemophelia day. In Kampala, there will be a blood donation drive at City Square organized by Nakasero Blood bank in partnership with Haemophilia Foundation of Uganda.

There will be screening of new patients in the West-Nile region and people intending to participate are expected to converge at Nebbi Hospital.

There will be a medical workshop about the disease in the same region.

TV and radio talk shows have also been organised on this day in different languages in some parts of the country, and all these efforts are geared towards increasing awareness.

Screening tests and treatment of the disease

Diagnosis involves knowing the history of the patient by asking questions such as when the bleeding started, if the person has had episodes of prolonged bleeding, if they have ever experienced dark and big patches after an injection. For women, doctors ask if they always experience heavy menstrual periods which are prolonged.

Diagnosis includes screening tests and clotting factor tests. Screening tests are blood tests that show if the blood is clotting properly. Clotting factor tests are required to diagnose a bleeding disorder.

This blood test shows the type of haemophilia one has and the severity. Then the doctors can prescribe treatment.

Dr Henry Ddungu, a haematologist at Uganda Cancer Institute, says treatment focuses on managing the bleeding disorder and regular replacement of the specific factor that is reduced or lacking.

He, however, notices that apart from medications, patients with haemophilia also need physical therapy which is aimed at maintaining range of motion (ROM) and strengthen all joints and prevent or diminish disability.

Additionally, Kasirye explains that the factors given to these patients are in two forms.

The first form is the recombinant factors also known as artificially prepared formulation factors and this is the current way of managing the disease.

Haemophilia is also graded from mild, moderate to severe. The grade is determined by the amount of the factor in the blood.

In mild haemophilia, a child makes in between 5-10% of the normal amount of factor, those with 1-5% active factor activity have moderate haemophilia and individuals with less than 1% of active factor activity are classified as having severe haemophilia.

"These patients are given the factors periodically to maintain the acceptable level, and our target is to raise the factor to at least 50% to 100%.

Every time we give factors, you create near normal situation before going for events such as operation," says Kasirye.

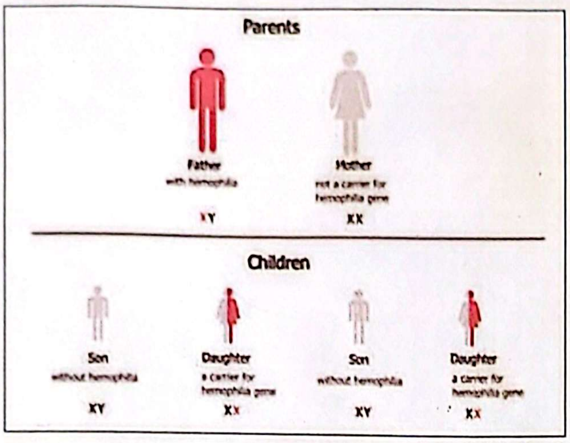
He further explains that the patients are put under the chronic care programme at Mulago Hospital and those with severe haemophilia are required to visit the facility weekly or twice a month.

Those with mild haemophilia are required to visit the facility once in a month or every two months to monitor their progress and get booster factor dozes.

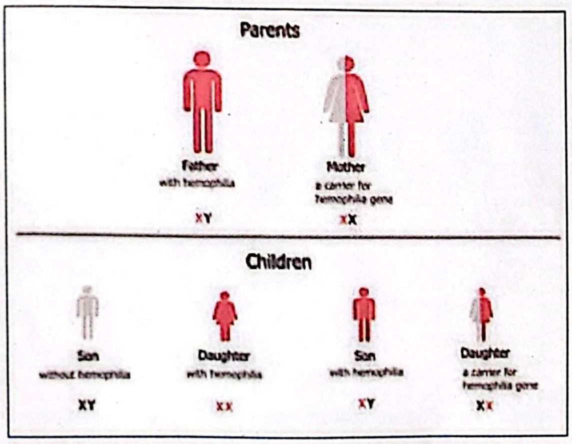
Dr Phillip Kasirye, a paediatrician at Mulago, points out that the artificial/ manufactured factors are made with protein structures, and though they are the safest, they are expensive. The second option is the natural factors produced by the body. This is got from blood products such as frozen plasma which has factor 8 and 9.

This plasma is obtained from Nakasero Blood Bank.

Haemophilia is a man-only disease



The above graph shows the odds when the father has haemophilia. The one below shows what happens when both parents are carriers.



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is according to referrals made to Mulago. Out of 100% of children with the disease, only 2% have been diagnosed. This means 98% remain under diagnosed and unidentified and their whereabouts are unknown.

Kasirye points out that since Mulago Hospital started partnering with the Haemophilia Foundation of Uganda a few years ago to organise medical camps, more patients have been diagnosed.

He explains that the largest challenge is that most Ugandans do not know about this disease.

Kasirye recommends continuous sensitisation about the disease.

He disclosed that Mulago is just developing a registry of patients with haemophilia. So far 150 have been identified, with more staff being trained to handle such patients.

Kasirye says the main management approach lies primarily in educating parents about the disease.

"The most critical stage is when a child with haemophilia starts crawling. Just like any other child, these children are also very active at this stage, they want to move around the entire house and would want to play with different objects."

It is advisable for parents/guardians to keep away sharp objects in the house. The experts also say children with haemophilia should not be engaged in strenuous activities such as bicycle riding or sports for a long time, as these can predispose them to falls. "In case the child is playing with peers, a parent should talk to them so that they do not push the child, or bruise him."

Kasirye says as children with this condition grow, a parent should counsel them in an age-appropriate way, tell them about the condition and things that are likely to predispose them to bleeding.

RICE management

A parent may try as much as possible to keep the child grounded so that no harm comes to them, but children like to explore and in the process may harm themselves and bleed.

Kasirye says the RICE (rest, ice, compression and elevation) approach is recommended. RICE is a treatment many health care professionals recommend for joint bleeds, it also reduces swelling and tissue damage when used together with clotting factor concentrates.

Applying ice helps control swellings and reduce pain. However, the ice should not be applied for too long as this can cause muscle weakness or kill the cells.

Compression means applying pressure on the cut area which helps stop the bleeding. Elevate and rest the injured area. If moving the injured area causes pain, this is the body's way of saying stop.

Prevention

Both Dr Henry Ddungu, a haematologist and Kasirye say the only prevention is through genetic counselling. Ddungu says couples should be screened, and if one has the disease, they can make informed decision to get married or not.

Kasirye strongly advises women who are carriers of the gene not to marry a man with haemophilia, adding that by doing this, the couple stands high chances of giving birth to children with this disease.

Where to get help in Uganda

Kisakye leads awareness team

At the moment, the Haemophilia Association of Uganda is the only organisation in the county that handles patients of haemophilia and their family members.

The executive director of the association, Agnes Mavis Kisakye, says they are engaged in creating awareness about the disease through community outreach.

The association also educates family members and parents on how to take care of patients. They also empower the youth with the condition so that they are able to accept the situation and live productive lives.