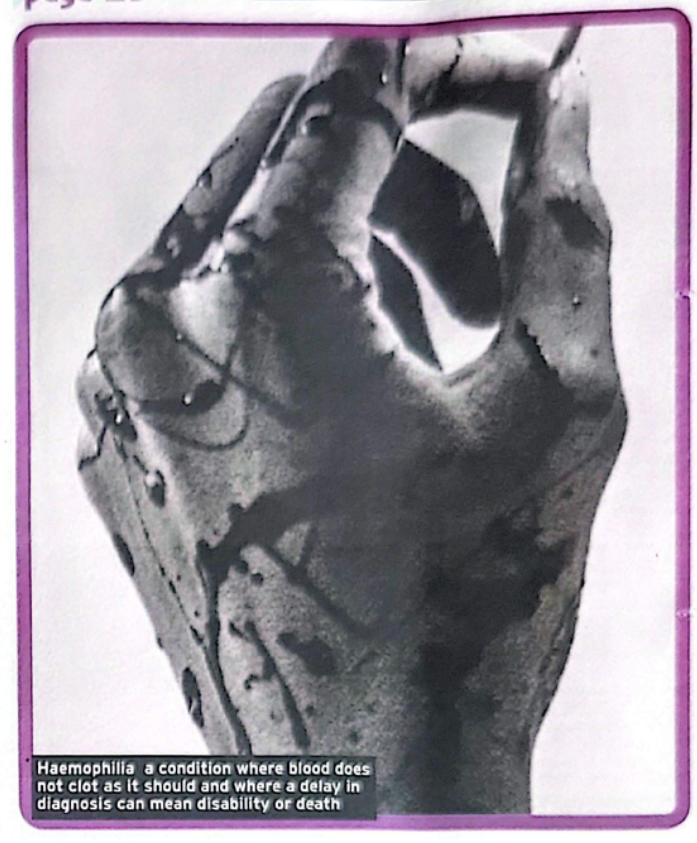
HAEMOPHILIA BURDEN: A CALL FOR DIAGNOSIS

As the world marked World Haemophilia Day on April 17 under the theme Diagnosis: First Step to Care, attention turned to a reality affecting thousands: a condition where blood does not clot as it should and where a delay in diagnosis can mean disability or death, writes **Jackie Nalubwama**



Haemophilia a condition where blood does not clot as it should and where a delay in diagnosis can mean disability or death

On most days, a small cut is nothing more than a fleeting inconvenience – wipe the blood away, wait a moment and life goes on. But for people living with haemophilia, that same cut can be dangerous, even life-threatening.

As the world marked World Haemophilia Day on April 17, under the theme *Diagnosis: First Step to Care*, attention turned to a reality affecting thousands: A condition where blood does not clot as it should and where a delay in diagnosis can mean disability or death.

According to the World Federation of Haemophilia (WFH) website, it is estimated "that over three-quarters of the population of people with haemophilia are undiagnosed and an even more significant gap also exists for other bleeding disorders."

WFH is a non-profit organisation dedicated to improving and sustaining care for people with inherited bleeding disorders worldwide.

In an in-depth interview with Dr Peter Wasswa, a haematologist at Mulago Hospital, explained that haemophilia is a hereditary disease. He revealed that one in 5,000 males worldwide suffer from haemophilia.

"Based on our population and assuming most survive to adulthood, Uganda is estimated to have over 4,000 patients," he said.

The gene that causes the disease is located on the X chromosome.

"Chromosomes come in pairs with men having XY and females having XX. Because females have two X chromosomes, having one defective X chromosome does not lead to disease, because the normal X chromosome will cause factor production. As such females hardly ever develop haemophilia. Those with one normal and one deficient X chromosome are called carriers and give birth to boys with haemophilia (a 59% chance) or girls who are carriers themselves (a 50% chance). The males have XY, so those born with an abnormal X chromosome will develop haemophilia," he explained.

SIGNS AND SYMPTOMS

Dr Wasswa said the hallmark feature of haemophilia is abnormal bleeding, which is excessive and prolonged relative to the injury.

"For the most severe types of haemophilia, excessive bleeding occurs after minimal trauma, often spontaneously. For example, a patient may present day after circumcision or a tooth extraction in which bleeding may be severe enough to require blood transfusion and may last days," he said.

He also noted internal bleeding in the joints as another sign, which is a common happening causing painful, stiff or swollen joints.

"Pain and swelling are the most common features. For example, joint bleeds [the most common complaint] manifest as joint pain, joint stiffness and swelling. Muscle bleeds manifest as swelling and pain. Brain bleeds often present as severe headache, sometimes with convulsions or loss of consciousness," he explained. "In essence, the symptoms depend on the bleeding site, with pain being a common denominator."

The bleeding can become severe. "Joint bleeds can lead to crippling. When bleeding occurs within the head, it can cause headache, seizures or even loss of consciousness," Dr Wasswa said.

The key signs of haemophilia are easy bruising and prolonged bleeding even after a simple procedure like circumcision or getting an injection, said Dr Phillip Kasirye, a haematologist at Mulago Hospital.

And if diagnosed late, Dr Kasirye said it can result in deformity or death.

COSTLY TREATMENT

Although some have an exaggerated fear of the condition, Dr Wasswa said it is treatable.

"The treatment is with replacement of the deficient factor, that is, factor 8 and factor 9 for haemophilia A and B, respectively. The treatment is administered by an injection in the vein. This is done whenever the patient develops bleeding symptoms, has been injured or is to undertake procedure that causes bleeding, such as surgery or tooth extraction. This is called episodic treatment," he explained.

Although the episodic treatment is helpful, Dr Wasswa said it is a reactive approach and not a preventative one.

Worse, Dr Wasswa said some patients may develop silent bleeding in the joints, which isn't recognised and treated, leading to joint damage.

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