**Encouraging genetic counselling**

Dr Munira Borhany

Thursday, Jun 02, 2022

Due to a lack of awareness among the people as well as doctors – who are also inadequately trained to manage them – haemophilia and other inherited bleeding disorder patients continue to suffer. The government must step up and help put an end to the suffering by allocating a sufficient budget towards their medical expenses, including the provision of lifesaving medicines, such as anti-haemophilia injections.

Haemophilia is an inherited disorder characterized by prolonged bleeding, either spontaneously or after injury from any part of the body or surgery. Blood contains many proteins called clotting factors that can help stop bleeding. People with haemophilia have deficiency or reduced activity of clotting factor, factor VIII (hemophilia A) or factor IX (hemophilia B).

The severity of haemophilia is determined by the amount of factor levels in the blood. The disease is classified as severe, moderate, or mild. People with mild haemophilia rarely bleed unless after trauma or invasive procedures. Frequency and severity of bleeding is greatest in severe haemophilia patients. Thus, the lower the amount of the factor, the more likely it is that bleeding will occur which can lead to serious health problems. Spontaneous bleeding includes excessive bleeding from ‘everyday’ injuries. Easy bruising in childhood, bleeding into the joints and muscles. Bleeding into other parts of the body such as the nose, gums, gut, kidneys, and head. Excessive bleeding during or after surgery even simple procedures can result in excessive bleeding e.g. tooth extraction and circumcision.

Haemophilia A is more common than Haemophilia B, representing 80-85 per cent of the total haemophilia population. These two diseases are clinically alike, but the treatment is different and includes replacement of missing clotting factor concentrate as soon as possible to treat or prevent bleeding. According to the World Federation of Hemophilia (WFH), the prevalence of affected hemophilia patients worldwide is 21 in every 100,000 male births.

In Sindh, affected haemophilia patients are 5234, out of which 766 haemophiliacs (out of total 900 bleeding disorders) are registered at the Hemophilia Welfare Society, Karachi (HWSK). This leaves 4332 unidentified due to lack of awareness and high cost of basic diagnostic tests such as complete blood counts, coagulation tests and factor levels. In the developing world, where the majority of haemophiliacs live, awareness of this disease and its management is poorly done. This leads to a significant cause of morbidity and mortality and is responsible for psychological, social and economical stress to patients and their families.

The standard of care for this disease is treatment with clotting factor concentrates (Anti-Hemophiliac injections) to prevent and treat bleeding. Unfortunately, in Pakistan due to non-inclusion of haemophilia treatment in the provincial and federal health budgets, it is not accessible for haemophiliacs and they are treated with alternative treatments like blood plasma therapy from public-private hospitals & blood banks for many decades. Due to unsafe sale of blood there is a high risk of transfusion transmitted infections such as hepatitis B, hepatitis C, HIV and because of this alarming number of hemophilia patients are positive for these infections.

Recently, in collaboration with the Sindh Blood Transfusion Authority (SBTA) and the Sindh health department’s programme for communicable diseases, hemophilia patients at HWSK were tested for these viruses. Unfortunately, 50 per cent of the patients tested positive for Hepatitis C infection, five per cent for Hepatitis B and one per cent for HIV.

Therefore, haemophilia personnel deserve proper diagnosis and treatment facilities. If these are not made available immediately, the affected and the unidentified are at a mortality risk due to constant (internal/external) joint bleeds, intracranial bleeds, gastro-intestinal bleeds which will lead to complete physical disabilities. In the current scenario, unfortunately all the patients above 12 years are heading towards physical deformities rapidly and due to this they are unable to acquire education. This burden gives birth to additional psychosocial issues not only in them but also in people close to them.

In recent years, prophylactic therapy which aims to prevent future bleeds in particular joint bleeds has become the standard of care. The World Federation of Hemophilia (WFH) guidelines recommend prophylaxis to prevent bleed and joint destruction and preserve normal musculoskeletal function. Moreover, it is advisable to give it to all patients before performing any activities associated with an increased risk of trauma.

Prophylaxis should be a state-of-art treatment. Despite the guidelines, prophylaxis has not been universally adopted and in Pakistan particularly. Medical, psycho-social, cost controversies and affordability issues limit the implementation of prophylaxis and the majority of patients are treated by on-demand administration of factor concentrates to stop bleeding. At the HWSK, 25 children less than 10 years have been put on prophylaxis treatment with long-acting factor concentrate to prevent bleeding. Furthermore, 36 severe haemophilia A patients are prophylactically treated with new modern medicine Emicizumab therapy in collaboration with the WFH humanitarian aid programme. These prophylactic treatments have markedly improved the quality of life of these patients.

In order to decrease the haemophilia mortality and disability rate, there is a dire need of public-private partnership between provincial governments, organizations that work on haemophilia and the World Federation of Hemophilia. This will make the upcoming haemophilia generation a functional part of society rather than being seen as a burden, like the previous generations. Furthermore, awareness, education and genetic counselling is needed to prevent the spread of such common occurrences of these bleeding disorders in the community.

The writer, a consultant haematologist, heads the Hemophilia Welfare Society, Karachi (HWSK).