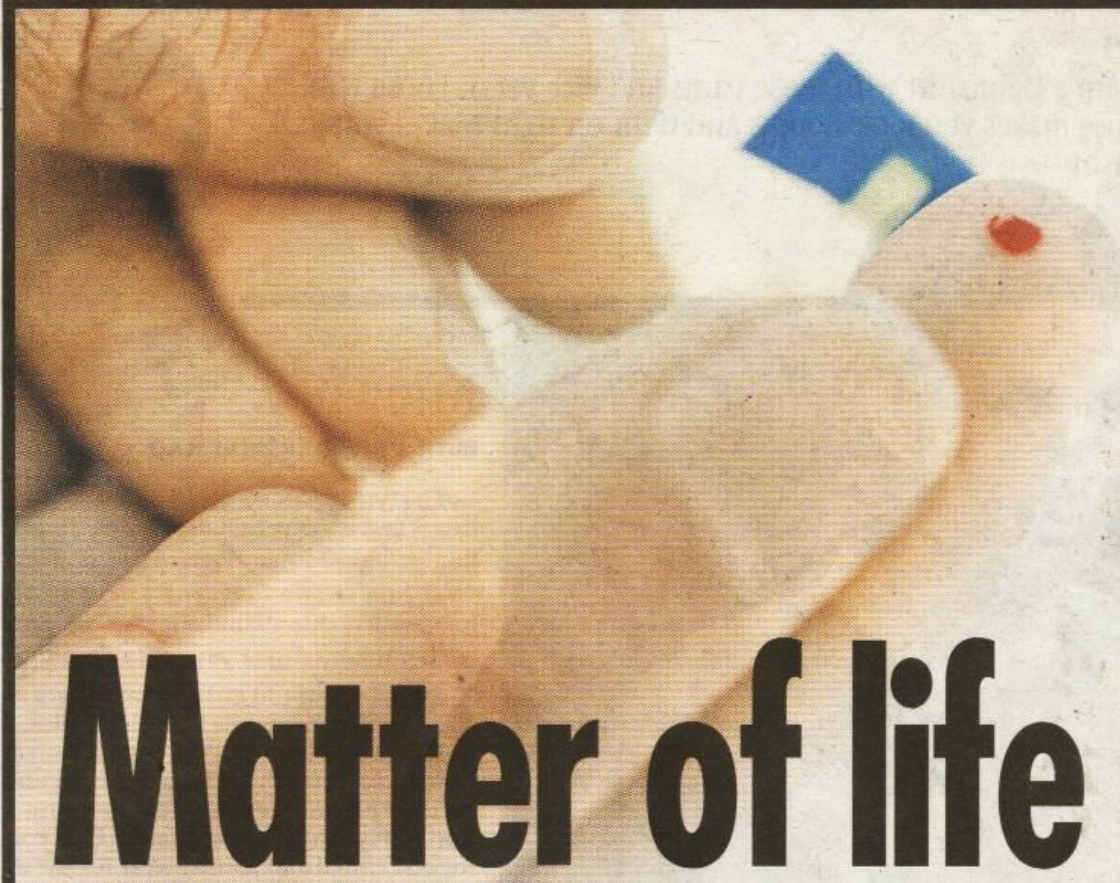

Matter of life

On World Haemophilia day, Kiran Mehta speaks to **Dr Abhay Bhave** on the illness

Understanding Haemophilia

Haemophilia is a bleeding disorder wherein a person presents has excessive bleeding spontaneously or in response to a minor trauma. Largely an inherited condition, haemophilia can sometimes be acquired as a result of the body's response to another ailment such as malignancy or autoimmune disorders of the blood.

There are two basic types of haemophilia - haemophilia A and B. Heamophilia A occurs as a result of factor viii (a type of protein) deficiency. Whereas, haemophilia B occurs as a result of factor ix deficiency. The deficiency occurs due to chromosomal defect in a specific portion of the chromosome that is responsible for the production of factor viii or ix protein.

Men beware!

Since haemophilia is an X-linked recessive disorder, it largely affects men. However, a woman can be a carrier of the disorder and in extremely rare cases she herself may manifest symptoms of the disorder.

Am I suffering from Haemophilia?

Affected children present with bleeding at circumcision, bleeding following intramuscular immunisation, excessive bruising or in rare cases with bleeding in the brain. Mothers may note easy bruising or blood collection on the lip (haematoma) of the child as soon as he is old enough to crawl which is when children are prone to minor injuries.

Patients with milder forms of haemophilia may present no symptoms until they have been challenged either by a physically traumatic event or excessive bleeding may be noted during a surgical activity. Typically the patient does not bleed immediately following the trauma, instead, bleeding occurs after a gap of a few hours.

The most common cause of distress is bleeding in closed joint spaces (hemarthrosis). Other symptoms may include epistaxis (bleeding from the nose, unrelated to weather) and excessive bleeding during a tonsillar or dental surgery.

Going in for diagnosis

A confirmed diagnosis can be made through a simple blood test referred to as the blood coagulation test.

Time factor

The type of haemophilia, whether mild, moderate or severe does not change with ageing. But the number of bleeding manifestations will vary depending on the lifestyle of the patient.

Danger to life?

If the bleeding is in a closed space such as the brain, it can lead to death. Excessive bleeding during surgery in an undiagnosed case can also lead to a life threatening disorder.

The treatment

During an emergency haemophilia is treated by injecting the patient with commercially available factor VIII or IX concentrates. Patients can purchase the concentrates and store them in case of an emergency. Since this a costly option, patients can approach the Heamophilia Federation of India (Mumbai Chapter), close to KEM hospital. And as members of this organisation they can procure the commercially available factors at more affordable rates. Patients can also opt for prenatal diagnosis if there is a family history. For those already affected by the condition, genetic counselling is advisable wherein they are advised to alter their lifestyles which brings down the incidence of the disorder and thereby improves the quality of the patient's life.

Another option being researched is gene therapy. Hemophilia A and B are ideal disease states to target since they are caused by mutations in single identified genes. So a slight increase in clotting factor levels in the blood can convert severe hemophilia into milder disease. Unfortunately, to date the promise of gene therapy and a cure for the hemophilia patient have not been realised.

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