DIAGNOSIS

If you or your child have been diagnosed with mild haemophilia, it is important to see a haematologist who specialises in bleeding disorders. In Australia these haematologists can be found at Haemophilia Centres or Services which are at some major hospitals. Talk to your doctor about a referral.

Haemophilia Centres have a team of doctors, nurses, social workers, counsellors and physiotherapists with expertise in providing treatment and care to people with haemophilia. They also have access to specialist laboratory and diagnostic testing and can give referrals to genetic testing and counselling services.

Although people are born with haemophilia, a person with mild haemophilia may not be diagnosed until they are older when they first experience unusual bleeding problems. This often occurs after an accident or injury or if the person has surgery or tooth extractions. Girls and women may become aware of bleeding problems when they begin to menstruate or after giving birth.

Haemophilia is usually diagnosed by assessing these three things:

- The person has a history of prolonged bleeding, for example, with injury or surgery, dental work or medical procedures
- Checking if there is a family history of haemophilia or bleeding problems
- Laboratory tests on a blood sample for a person's clotting factor levels.

Often further blood tests will be done when the person is reviewed at the Haemophilia Centre to confirm the diagnosis. Sometimes low factor VIII levels can also occur if the person has von Willebrand disorder, which is another type of inherited bleeding disorder. Identifying the people who should have additional testing requires specialised knowledge and expertise in bleeding disorders.

Family history

If someone is diagnosed with mild haemophilia, it is likely that other members of their family also have haemophilia or carry the gene. Diagnosis will also include checking the family history for bleeding problems. Other family members may also need to be tested for haemophilia.

If people who are having a child know that some people in their family have haemophilia, they should arrange with their Haemophilia Centre and obstetric teams to have a male baby tested soon after the baby is born to see whether he has haemophilia. Prior to delivery they may also choose to test for the sex of the baby. Genetic counselling is available to couples and families to discuss options.

This preparation can help with planning for a smooth and safe delivery and care of the newborn baby. Diagnosis at an early age enables both the family and health care teams to manage the child's health care plan safely. It also avoids discovering haemophilia under stressful conditions such as accident or surgery.

See also CARRYING THE HAEMOPHILIA GENE, page 40.

Other issues at diagnosis

Diagnosis with mild haemophilia may also raise other questions:

- Possible implications for employment, personal insurance cover and carrying the haemophilia gene
- Decisions, made in consultation with the Haemophilia Centre or Service, about which treatment options will be the most suitable.

The Haemophilia Centre team is available to help people deal with these issues and questions.

"I'm not sure when I first found out about my mild haemophilia – I think I was around 12-14 years old, because I started getting bleeds."

"When I was told, I didn't really understand the impact of my condition. I just wanted to be just like all the other boys and for a long time I was determined to be so and I largely ignored the condition making excuses for bruises and limps. I would strongly encourage parents to tell their child and siblings about the condition as soon as practicable - in words they understand."

"It's important for parents not to worry, but to educate yourself so you are in the best position to teach your children about their condition."

"My mother was not aware of her carrier status until I was about 12 or 13. My grandfather had gone in for surgery and he had had a bleeding episode - after the procedure the doctors asked him if he had haemophilia and he said yes. He had not told them prior to the procedure as he was ashamed of the condition. Once we found out he had haemophilia the family line was traced - my mother was a carrier, both myself and my brother had haemophilia."