Familial hypercholesterolemia

An estimated 4000 West Australians could be ticking time bombs, and up to 80 times more likely than the rest of the population to have a heart attack or develop premature heart disease because of a genetic disorder characterised by markedly elevated cholesterol levels.

Known as Familial hypercholesterolemia (FH), the disorder is considered a silent condition because many sufferers are not aware they have it until they develop angina or have a heart attack. Gerald Watts, head of the lipid disorders clinic at Royal Perth Hospital, said FH was a dangerous condition when left untreated, with half of its sufferers having heart attacks before the age of 50 for men and 60 for women.

Professor Watts said the gene mutation for FH only needed to be present in one parent for the disorder to be passed on to their children. Gene mutations in both parents made the disorder even more life threatening but this was very rare. Recent surveys had shown about one-third of people with FH also displayed other risk factors which increased their heart attack risk, including obesity, smoking, high blood pressure and diabetes, Professor Watts said.

About one in 20 people with premature heart disease was likely to also have the FH gene.

The best way to treat FH was to pick the condition up early with DNA testing in children and teenagers, usually after puberty in boys and after a girl's first period. Treatments for the condition included a healthy lifestyle and dietary intake and the use of statin drugs. "It's absolutely fundamental that young people with FH do not smoke nor become obese," he said. In severe cases, where FH patients carried the gene (homozygous FH) from both parents, those patients required intensive life-long treatment. Statin drugs were combined with regular apheresis, a process similar to dialysis, to remove cholesterol-containing particles from the bloodstream.

FH has been a part of Simone Poor's life since she was only four years old, diagnosed with the condition when she started developing cholesterol deposits on her hands, elbows, knees and ankles.

Ms Poor is one of only six people in WA to carry the FH gene inherited from both parents.

Connie Clarke

For more information about FH, contact the Genetic Support Council WA on 9485 8999 or visit www.familialhypercholesterolaemiasupportwa.websyte.com.au

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