An impressive new consensus paper prepared by the European Atherosclerosis Society (EAS) has addressed the issue of under-diagnosis and under-treatment of heterozygous FH (1). This EAS Consensus Paper estimates that there are between 14 and 34 million individuals with FH worldwide. It notes that in many countries, less than 1% of those with the condition have been diagnosed. And even in countries where a greater proportion of those with the disorder are diagnosed, the paper concludes that a disturbing number of those with FH are not being treated with cholesterol-lowering therapy.

Familial hypercholesterolemia (FH) is a genetic disorder characterized by markedly elevated levels of low-density lipoprotein (LDL) cholesterol and is well known to cause premature cardiovascular disease (CVD). Most people with FH have a mutation of the LDL receptor gene resulting in reduced activity of the LDL receptor, the cell surface protein that binds LDL particles in a process that removes them from the plasma. This condition has been widely investigated and its molecular mechanisms are well understood.

People who are homozygous for this condition have a virtual absence of active LDL receptors and have plasma cholesterol levels as high as 30 mmol/L (1200 mg/dL). Such people develop CVD very early in life and, if untreated, usually die before age 20. While homozygous FH has a prevalence of only about 1 in 500,000 to 1 in 1,000,000, those who are heterozygous for the condition represent somewhere between 1 in 200 and 1 in 500 of the population worldwide. This translates into a very large number of people with the condition.

People with heterozygous FH have plasma cholesterol levels as high as 15 mmol/L (600 mg/dL) and, if untreated, typically have a major CVD event before age 60. However, if they are diagnosed, people with heterozygous FH can be treated with cholesterol-lowering medication that has been shown to slow (or even reverse) the development of atherosclerosis and markedly reduce the risk of having a clinical CVD event.

It is thus alarming to note that the vast majority of people who have heterozygous FH do not know that they have the condition.

The EAS Consensus Paper is comprehensive and evidence based. It is aimed at cardiologists, endocrinologists, internists, paediatricians, general practitioners, clinical biochemists, public health practitioners, health service planners, other health professionals, and health care providers worldwide.

In my view, the paper should be compulsory reading for all who are committed to reducing the worldwide burden of atherosclerosis and its associated cardiovascular disease.

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