

'Silent' high cholesterol condition diagnosed too late and undertreated

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A new report has highlighted the late diagnoses and undertreatment of a genetic condition which causes cholesterol levels to soar.

Familial hypercholesterolaemia (FH) is an inherited condition affecting as many as 1 in 300 people around the world, thus it may affect more than 25 million people around the world.

It can affect otherwise healthy individuals, irrespective of their lifestyle, causing them to have higher than normal levels of LDL cholesterol from birth.

This puts them at greater risk of cardiovascular disease if they are not treated with cholesterol-lowering drugs, such as statins.

In the most accurate global snapshot to date of FH and how it is managed, investigators led by researchers at Imperial College London have highlighted how the condition is being diagnosed too late in life, that greater use of intensive cholesterol-lowering drugs is needed, and that the diagnosis and treatment for women is falling behind that of men.

Their findings, published today in *The Lancet*, are the first to come from a global registry of more than 42,000 adult patients with FH from 56 countries, developed by the European Atherosclerosis Society Familial Hypercholesterolaemia Studies Collaboration (FHSC).

Dr. Antonio Vallejo-Vaz, from Imperial's School of Public Health and lead author of the report, said: "As an inherited condition, FH is diagnosed too late, on average in the mid-40s in the FHSC adult population, meaning that many years elapse before patients are identified and treatment is started.

"Late diagnosis also potentially misses out on opportunities to address other cardiovascular risk factors which become more prevalent with increasing age. Identification of FH must be improved to detect those affected much earlier."

Delayed diagnosis

Among their findings, researchers found that FH diagnosis is usually delayed, with less than half of [adult patients](#) (about 40%) under 40 years of age when diagnosed.

The registry shows that among approximately 30,000 adults for whom there is data, the median age of [diagnosis](#) was 44.4 years, with one in six already having heart disease at entry to the registry.

Among those patients on lipid-lowering therapy (59.9% of registry), most were on a [statin](#) (81%).

However, few were on the highest doses of statins (which may deliver the greatest benefit for FH) and only about one in five were on a combination of lipid-lowering therapies.

Compared with men, women were less likely to receive the highest doses of statins or combination therapy, despite having higher LDL-C levels from age 50 years.

Overall, less than 3% of patients on treatment attained LDL-C levels below 1.8 mmol/L (

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