'Silent' high cholesterol more common than thought, warns leading global report

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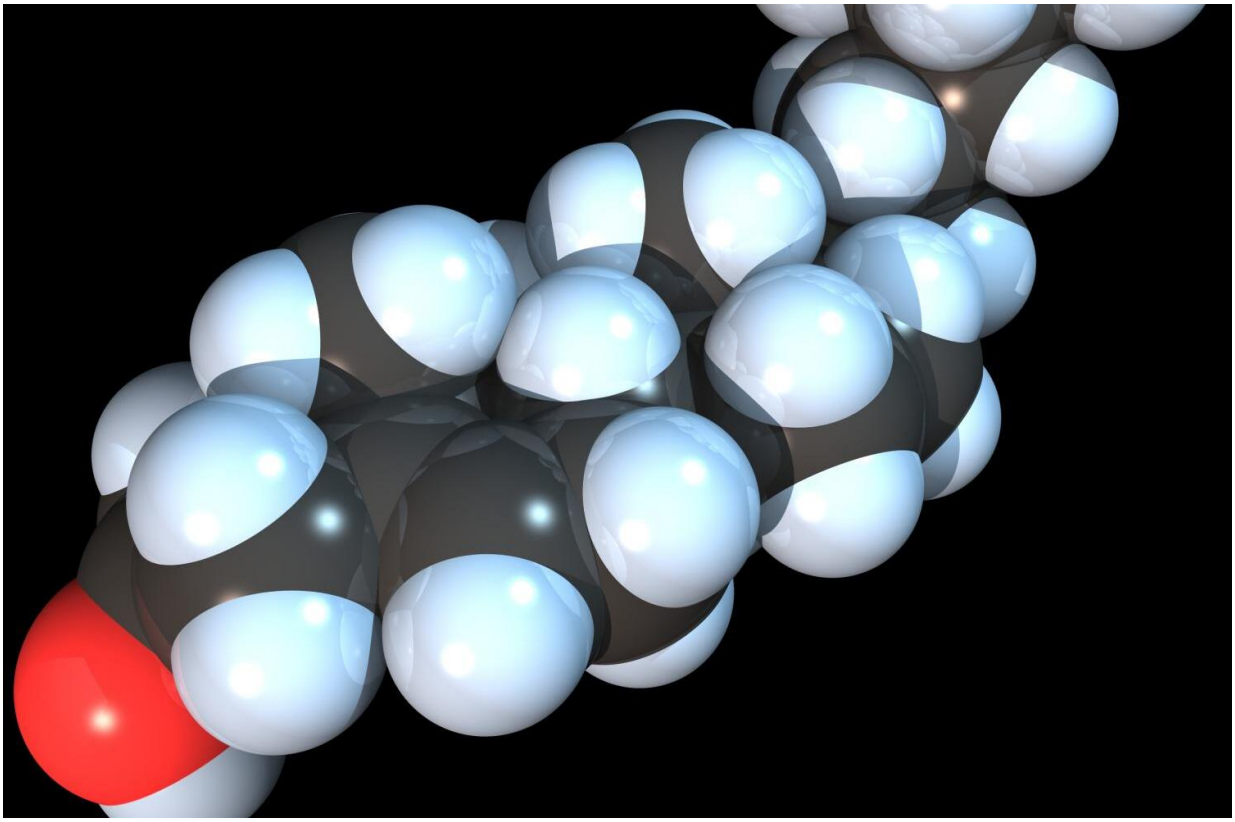

Space-filling model of the Cholesterol molecule. Credit: RedAndr/Wikipedia

Improving the knowledge of a genetic condition which causes cholesterol levels to soar could lead to fewer people dying from heart disease.

The findings come from the largest report to date on the global status of familial hypercholesterolaemia (FH), a hidden killer which can affect otherwise healthy individuals irrespective of their lifestyle.

FH is an inherited condition which causes people to have higher than normal levels of LDL cholesterol, commonly referred to as 'bad' cholesterol, leaving them at greater risk of cardiovascular disease if the condition is untreated with cholesterol-lowering drugs, such as statins.

Even if a person has a healthy lifestyle, having FH means they can still have a much higher risk of blocked arteries which reduce the blood flow to the heart and are typically associated with eating too much fatty food, smoking and an inactive lifestyle.

Underestimated prevalence

The condition was previously though to affect an estimated 1 in 500 people, but in the last decade it has emerged that this figure has underestimated the true incidence by more than half, with researchers estimating FH may affect as many as 1 in every 200-300 people worldwide.

Now, an international consortium (European Atherosclerosis Society Familial Hypercholesterolaemia Studies Collaboration – FHSC) led by researchers at Imperial College London has produced the most accurate picture to date of the global status of the condition, and how it is managed.

The report, published in a special edition of the journal Atherosclerosis, reveal an increased prevalence of FH in the global population, as well as identifying a lack of information on the condition and gaps in its screening and treatment.

Professor Kausik Ray, Chair in public health (clinical) from Imperial's School of Public Health and chair of the FHSC, who led the study, said: "For the first time, we have been able to show the true scale of FH in terms of its global prevalence and how well it is being managed.

"Many healthy people may be living their lives unaware they have an underlying genetic condition which puts them at significantly higher risk of heart attacks and cardiovascular disease, independent of their lifestyle factors."

Since 2015, lead investigators from around 70 countries have joined the FHSC, creating an international network and the only global registry on FH – including more than 11,000 patients worldwide so far.

In its report, published this week, the investigators gathered data from a total of 63 countries around the world, focusing on three aspects: available information on the prevalence of FH, programmes and initiatives for FH screening, and the management of the condition in each of the countries.

Information lacking

Among the group's key findings are that information on FH prevalence is lacking in most countries, with low rates of FH identification reported universally; education programmes to improve FH awareness and knowledge are a recognised priority.

The investigators found that only some countries have adequate genetic screening capabilities in place, with many programmes being supported locally or regionally rather than at the national level, and frequently lacking support.

Their study showed that high-intensity statins – the standard treatment

for FH – were available in all nations surveyed, but that under-treatment was common, and that the cost of medications were not universally covered by healthcare systems.

In addition to the baseline statin treatment, the investigators also looked at the availability of three additional therapies for FH: ezetimibe, PCSK9 inhibitors and lipoprotein apheresis.

They reveal that ezetimibe was available in all but four of the studied countries, while PCSK9 inhibitors were available in almost two-thirds of countries. Lipoprotein apheresis – which physically removes excess from the patient's blood in a process similar to dialysis – was available in approximately 60 per cent of countries surveyed (and frequently limited to a few centres of reference).

In the UK, it is estimated that fewer than 20 per cent of cases of FH are diagnosed, with access to genetic testing limited, but all three classes of drug treatment were available.

Commenting on the findings, Dr. Martina De Marco, from the School of Public Health and co-first author of the paper, said: "This report has shown clear gaps in the knowledge and available information on FH around the world. Improving knowledge of the condition universally, including understanding the differences between different regions, as well as improving access to treatments and funding of health programmes to tackle FH, are key priorities."

Dr. Antonio J. Vallejo-Vaz, from the School of Public Health and co-first author of the paper, added: "This study identifies a clear need to improve screening for FH, as well as screening for family members of those affected by the condition. The earlier we can identify patients, the earlier we can intervene – either through offering advice on dietary and lifestyle changes, or through prescribing preventative medication, such

as statins and PCSK9 inhibitors – to help lower their cholesterol levels and reduce their risk of cardiovascular disease in the long term. The findings from this survey may ultimately help inform public health policies on FH."

Professor Lale Tokgozoglu, President of the European Atherosclerosis Society, commented: "FHSC is the largest report to date on the global status of familial hypercholesterolaemia and will provide a huge amount of information on familiar hypercholesterolemia.

"These data will help implement best practises throughout many countries. Understanding the disease will help us fight it better. National societies will work to improve outcomes in patients as well as help to build national heart health policies."

More information: Antonio J. Vallejo-Vaz et al. Overview of the current status of familial hypercholesterolaemia care in over 60 countries - The EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC), *Atherosclerosis* (2018). DOI: 10.1016/j.atherosclerosis.2018.08.051

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