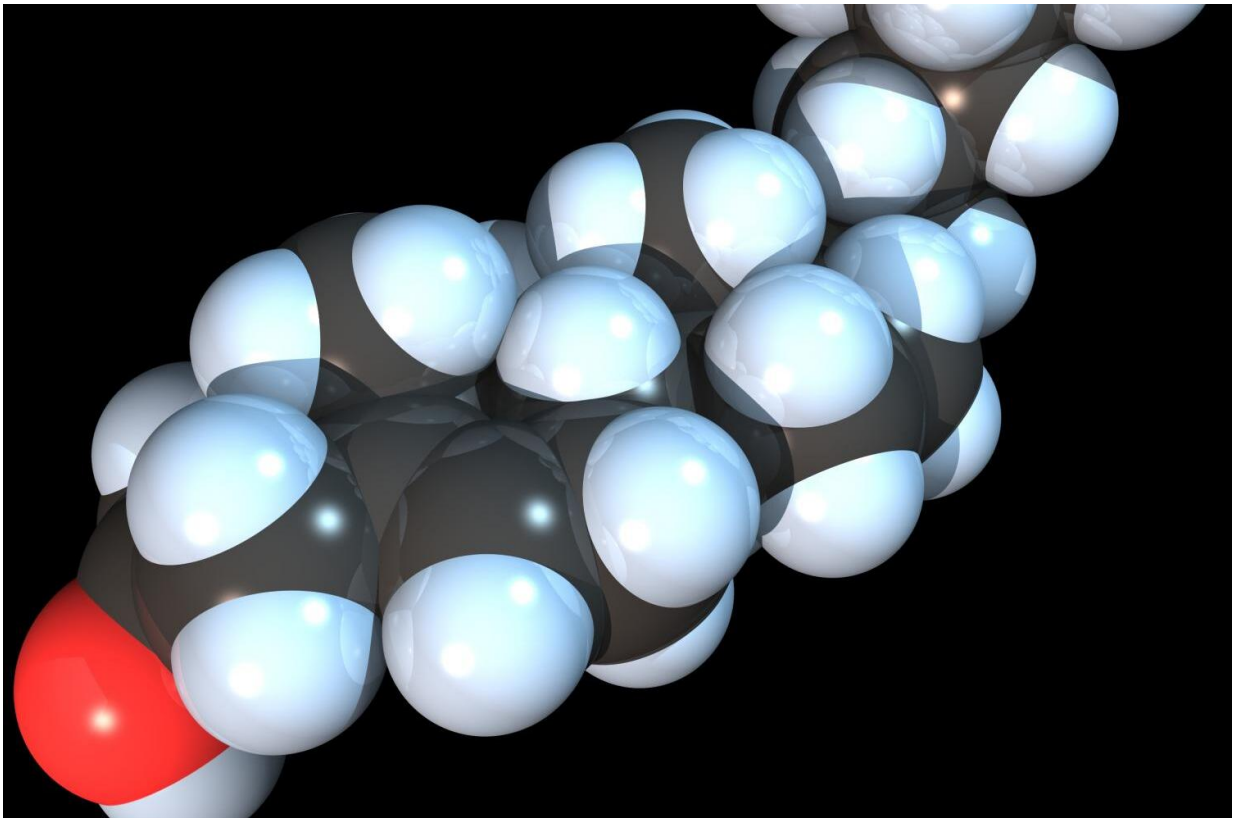
New gene for familial high cholesterol

May 12 2016



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

New research from Denmark reveals the gene that explains one quarter of all familial hypercholesterolemia with very high blood cholesterol. Familial hypercholesterolemia is the most common genetic disorder leading to premature death, found in 1 in 200 people.

A research group lead by Clinical Professor Borge G. Nordestgaard has found that cholesterol-containing lipoprotein(a) is the cause of one quarter of all diagnoses of familial hypercholesterolemia. High levels of this genetically determined lipoprotein in the blood is already known to cause heart attacks.

"Among 46,200 individuals in the general population, individuals with a diagnosis of familial hypercholesterolemia have higher levels of lipoprotein(a) in their blood than individuals without the diagnosis," says principal investigator Dr. Anne Langsted, Herlev Hospital, Copenhagen University Hospital, Denmark.

The research has just been published in *The Lancet Diabetes & Endocrinology*.

High risk of heart attack

The study also reveals that high levels of lipoprotein(a) in the blood adds to the already very high risk of suffering a heart attack for people with familial hypercholesterolemia."We find that individuals with familial hypercholesterolemia and high levels of lipoprotein(a) are five times more likely to suffer a heart attack than individuals without these two conditions," adds Anne Langsted.

"Our results suggest that all individuals with familial hypercholesterolemia should have their lipoprotein(a) concentrations measured in order to identify those with the highest concentrations and therefore also the highest risk of suffering a heart attack," says senior author Borge G. Nordestgaard, University of Copenhagen and Copenhagen University Hospital.

"Worldwide, familial hypercholesterolemia as well as high lipoprotein(a) levels are grossly underdiagnosed and undertreated. Our findings will

help identify the individuals with the highest risk of suffering a heart attack and hopefully facilitate better preventive treatment for these extremely high risk individuals," adds Nordestgaard.

The diagnosis of familial hypercholesterolemia is based on different clinical signs such as elevated cholesterol, relatives with early heart attack and a personal history of heart attack. Genetic testing can also confirm the diagnosis.

Provided by University of Copenhagen