Screening for genetic high cholesterol could help patients and families avoid heart attack

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Genetic high cholesterol is underdiagnosed and undertreated, according to research presented today at ESC Congress 2019 together with the World Congress of Cardiology. Screening could identify patients and family members affected by the condition so that lifestyle changes and treatments can be started to prevent heart attack and stroke.

Heterozygous familial hypercholesterolaemia (FH) is a life-threatening genetic condition linked with a high risk of premature cardiovascular disease, including heart attack and stroke. FH is one of the most common potentially fatal family disorders, with a prevalence estimated at 1/250 to 1/200, corresponding to 3.6 to 4.5 million individuals in Europe.

Patients with FH have high levels of "bad" cholesterol (low-density lipoprotein; LDL) due to a mutation in genes that clear cholesterol from the body. LDL particles accumulate in the blood and can ultimately build up in the coronary artery walls. Children of patients with heterozygous FH have a 50% chance of inheriting the disorder.

As LDL cholesterol levels are elevated as early as birth, the risk of heart attack in patients with FH is 10 to 13 times greater than that of the general population. Elevated LDL cholesterol plus family or personal history of early heart disease are key criteria for diagnosis, which may be confirmed by genetic testing. Management of FH includes a healthy lifestyle and medication.

This study examined the frequency of FH in the RICO survey, a large French database of patients hospitalised for a heart attack between 2011 and 2017. The researchers determined whether patients had FH using LDL cholesterol levels and family or personal history of premature coronary artery disease. Treatments, patient characteristics, and severity of coronary artery disease were compared between patients with and without FH.

Among the 11,624 patients with heart attack, FH was not rare (2.1%). When compared to patients without FH, those with FH were 20 years younger (71 versus 51 years) and had more severe coronary lesions.

Senior author Professor Marianne Zeller of the University of Burgundy—Franche-Comté, Dijon, France said: "Taken together, the earlier onset and severe lesions show the aggressive nature of coronary artery disease in patients with familial hypercholesterolaemia."

Regarding lipid-lowering treatments, chronic statin treatment was used in 48% of FH patients and ezetimibe in 8%. "There was a dramatic underuse of drugs to reduce cholesterol levels," said Prof Zeller. "Nearly half had no chronic treatment before they had a heart attack and ended up in hospital. This indicates that at least half of patients with FH were probably unaware of their diagnosis and their heightened risk of heart disease, and also that their family members could unknowingly be affected."

"Systematic FH screening at the time of hospitalisation for a heart attack could identify these high-risk patients," she continued. "Screening is simple, mainly based on high levels of LDL cholesterol and history of early coronary artery disease (personal and/or family). Diagnosis can be confirmed by a genetic test, which is available in most European countries."

Compared to patients without FH, those with FH had a significantly lower rate of hypertension (59% versus 47%), diabetes (25% versus 17%) and prior stroke (8% versus 4%), but a higher prevalence of smoking (23% versus 56%) and a personal (15% versus 20%) or family history (18% versus 78%) of coronary artery disease.

She concluded: "Better identification of patients with FH is needed so that cholesterol lowering treatments can be started as well as recommended lifestyle modifications such as eat a healthy diet, be physically active and quit smoking. Once we know who the patients are, we can then look for relatives with the condition."

More information: The abstract "Prevalence and severity of coronary disease in patients with familial hypercholesterolemia hospitalized for an acute myocardial infarction: data from the RICO survey" will be presented during the session Coronary artery disease and comorbidities on Sunday 1 September at 14:30 to 15:40 in Agora 2 – Poster Area.

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