Adults with inherited high cholesterol are underdiagnosed and undertreated

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An estimated 1 in 500 people worldwide suffer from familial hypercholesterolaemia (FH), an inherited condition of extremely high cholesterol that is associated with premature heart disease and death. Despite this high prevalence, recent research funded by the Minneapolis Heart Institute Foundation (MHIF) confirms FH is underdiagnosed and undertreated. Thomas Knickelbine, MD, Preventive Cardiology Director at the Minneapolis Heart Institute at Abbott Northwestern Hospital, will present the results of research aimed at identifying just how prevalent FH underdiagnosis is at the American College of Cardiology (ACC) meeting in Washington, DC on March 30.

Dr. Knickelbine and a team of MHIF researchers analyzed the electronic health records (EHRs) of 391,166 consenting ambulatory patients seen at Allina Health locations between 2009 and 2012 to identify individuals who were at least 80% likely to have FH (determined by low-density lipoprotein [LDL or "bad" cholesterol"] levels and age). They discovered 841 patients (0.21% or 1 in ~465) who were likely to have FH—only 36 (4.3%) of them had been diagnosed with the condition. "Provider recognition of FH is extremely low," explains Dr. Knickelbine. "Our research shows we can effectively use EHR data to identify asymptomatic FH patients and improve early diagnosis and treatment of this high-risk condition."

In this large, ambulatory patient population, the researchers also found that FH was undertreated. Of the 841 patients likely to have FH, 64.8% were on statin medications, but an additional 25.8% were not currently

on a statin nor were they reported to be statin intolerant. In addition, only about one fourth of the identified FH group had achieved their NCEP LDL goal, and only 31% had been seen by a cardiovascular (CV) specialist.

Provided by Minneapolis Heart Institute Foundation