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8TH CARDIOVASCULAR NURSING & NURSE PRACTITIONERS MEETING

Familial hypercholesterolemia screening: Challenges and opportunities

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Familial hypercholesterolemia (FH) is an inherited disorder of lipid metabolism characterized by the severe elevation of low-density lipoprotein (LDL) particles in the bloodstream. FH affects every racial and ethnic group, appearing at a rate of 1/200 to 1/500 persons worldwide. Despite its status as the world's most common potentially deadly genetic illness, FH is highly under-diagnosed and undertreated in the United States and internationally. Without treatment FH increases the risk of cardiovascular disease to twenty times that of the general population. Cascade screening, a process of identifying affected family members of an FH index case, has been successfully executed in countries such as the Netherlands and the Czech Republic; however, the United States has fallen behind these nations in addressing FH on an epidemiological scale. Two primary factors have hindered successful screening for FH in the United States, and likely pose challenges in select countries internationally as well: disproportionately low funding allocations for FH research compared to its disease burden and strict privacy laws limiting contact with relatives of FH index cases. Cascade screening for FH has been repeatedly proven to be cost-effective in terms of quality-adjusted life years added, and serves as an opportunity to intervene early and halt the progression of atherosclerosis.

Biography

Lars Andersen has completed his Bachelor's degree in History and pre-medical coursework at Brown University in 2015. He has since performed research on disorders of lipid metabolism, focused on familial hypercholesterolemia, familial defective apolipoprotein B-100, and loss-of-function mutations in *APOC3*, at the Lancaster General Health Research Institute within the Penn Medicine network. He has authored several publications on familial hypercholesterolemia screening and identification strategies for local and general populations.

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