

Children with familial hypercholesterolemia are diagnosed late and under-treated

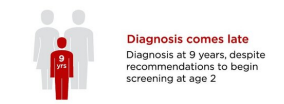
18 November 2020

CASCADE FH REGISTRY Children with Familial Hypercholesterolemia in the United States are Diagnosed Too Late and Undertreated

Enrollment data from 493 participants under 18 years of age in the Cascade Screening for Awareness and Detection of Familial Hypercholesterolemia (CASCADE FH) Registry in the United States.



Guidelines for children with Familial Hypercholesterolemia are not implemented consistently



GUIDELINES FOR CHILDREN

Screening
The American Academy of Pediatrics recommends that all children have a cholesterol test between the ages 9-11 and again between the ages of 17-21. Children from families with a history of early heart/vascular disease or familial hypercholesterolemia (FH) should have their cholesterol tested at the age of 2.

Treatment
U.S. and international guidelines recommend treatment with statins as early as age 8 and commencing by age 10 for children with FH, who are at high risk for early cardiovascular disease. Treatment goals for children with FH are LDL cholesterol under 130 mg/dL or 50% reduction in LDL cholesterol from highest pre-treated level.

Children with FH treated with statins had the greatest decrease in LDL-cholesterol



De Ferranti, Sarah D., et al. "Children with Heterozygous Familial Hypercholesterolemia in the United States: Data from the CASCADE FH Registry." *The Journal of Pediatrics*, 2020. <https://doi.org/10.1016/j.jpeds.2020.09.042>



Guidelines for children with familial hypercholesterolemia are not implemented consistently. Children with FH treated with statins had the greatest decrease in LDL-cholesterol. Credit: The FH Foundation

Children born with a common genetic condition, familial hypercholesterolemia (FH), are at increased risk for atherosclerotic cardiovascular disease (ASCVD) early in life. Despite long-standing national guidelines, cholesterol screening—for children at age 2 for those with a family history of heart disease and between the ages of nine to 11 for all children—is not being implemented.

New data from the FH Foundation's CASCADE FH

Registry highlights the gaps in care for [children](#) with FH, published online today in *The Journal of Pediatrics*. Among nearly 500 children and adolescents with FH, diagnosis occurred on average at nine years of age, seven years later than [cholesterol screening](#) guidelines.

"It is critical to identify children with [familial hypercholesterolemia](#) in order to prevent [cardiovascular disease](#) through early treatment intervention," said Sarah de Ferranti, MD, MPH, chief, division of ambulatory cardiology, Boston Children's Hospital, and CASCADE FH Registry principal investigator. "The U.S. guidelines are clear but there is a low level of implementing these guidelines that puts nearly 300,000 children at risk."

FH causes a high level of low-density lipoprotein cholesterol (LDL-C) from birth, that left untreated can cause early ASCVD including heart attacks and strokes. Treatment can begin as early as eight years old and is recommended to start no later than 10 years of age. One recent long-term study published in the *New England Journal of Medicine* demonstrated that initiating statins in childhood and continuing statins into adulthood led to a reduction of cardiovascular events as compared to their parents with the same genetic disorder.

Youth in the registry began lipid-lowering therapy on average at 11 years of age, with statins the most frequently reported medication (n=271; 56.2%). In addition, 23.5% of children eligible for treatment were not receiving therapy at enrollment. Only 39% of the youth achieved recommended LDL-C reduction according to U.S. and international guidelines (LDL-C

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