JSPCCS-AHA Joint Symposium

JSPCCS-AHA Joint Symposium (I-AHAJS)

Preventive medicine

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[I-AHAJS-01]Preventive Cardiology: Approaches to screening for lipid disorders in childhood

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Approximately 1 in 5 adolescents has an abnormal lipid level, with about 1% having significant confirmed hypercholesterolemia. Familial Hypercholesterolemia (FH) is a common genetic disorder, affecting one in every 200 to 500 people. Childhood lipid levels are associated with subclinical atherosclerosis in childhood and adulthood. Detecting and treating childhood lipid disorders, including FH, in youth may prevent future premature ASCVD. Screening for lipid disorders is therefore recommended by many cardiology and pediatrics guidelines, but despite its public health importance, is incompletely implemented in pediatric practice, genetic testing has not yet been integrated into general cholesterol screening practices, and the optimal age for first FH screening and the role of genetic and family FH screening have not been evaluated in the U.S. Low childhood screening rates are due in part to uncertainty about long-term efficacy of statins and lifestyle interventions, and concern about known and unknown adverse effects of statins. Pediatric lipid screening strategies include using genetic testing for FH mutations [e.g., LDL receptor (LDLR), apolipoprotein B (APOB), or proprotein convertase subtilisin/kexin type 9 (PCSK9) mutations], or "cascade" screening family members of FH cases. Cascade screening starting in childhood could be integrated into routine pediatric "well" visits (which children and parents attend), potentially allowing the initiation of preventive treatment of affected patients and their relatives years earlier than with adult screening. Newer machine learning methods could be applied to electronic health record data and patient population gene sequencing methods to make screening for lipid disorders during childhood more precise and efficient.