Screening for Familial Hypercholesterolemia: School Based Programs May Be Best Alternative

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Introduction

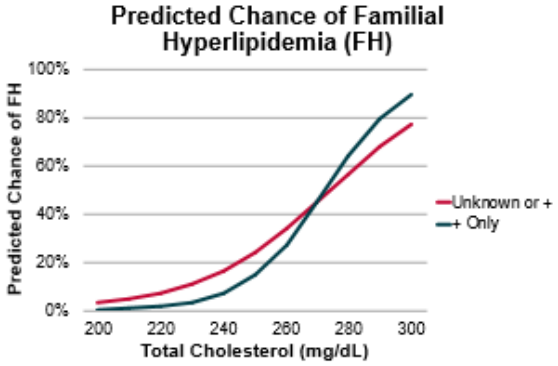
- Familial Hypercholesterolemia (FH) is a heterozygous autosomal dominant condition characterized by elevated cholesterol and premature development of cardiovascular (CV) disease affecting 1/200 - 1/500 individuals.
- Only 10% of the affected population in the United States have been identified and treated compared to 50-70% in Scandinavian countries identified through school-based screening programs.
- If left untreated, CV disease occurs approximately 10 years earlier which results in increased mortality, disability, and health care costs.

Methods

- The UCHealth Healthy Hearts and Minds Program (HHM) performs free biometric screenings on 5th, 7th, and 10th grade students in northern Colorado.
- HHM tested 10,991 5th grade students (9/2017 - 5/2022) and identified 424 students with total cholesterol levels ≥200 mg/dL (range 200-302).
- A buccal smear test (APOB, LDLR, PCSK9) was performed on 99 individuals with the highest total cholesterol levels prioritized (52% male, 10.6 years, 19.6 BMI).
- Binomial logistic regression model used for analysis.

Results

- Of the 99 students, six tested positive for FH (50% male, 10.3 years, 22.8 BMI) and five reported a variant of indeterminate significance (40% male, 11.3 years, 17.3 BMI).
- A sigmoidal association between total cholesterol and predicted chance of FH was identified with the inflection point at ~270 mg/dL.



These results further define the relation between total cholesterol and predicted chance of FH for a school-aged population where 245 mg/dL indicates a ~10% of testing positive for FH whereas 275 mg/dL has a ~55% chance.

Discussion

- The individual benefit of identifying FH early is the treatability of this condition. Early use of lipid lowering agents can make the CV disease risk equal to the general population.
- If elevated cholesterol is present in school aged children, genetic testing is warranted and treatment considered by age 8-10 years, especially if signs of abnormal vascular plaquing are identified.
- The multiplier effect documented through cascade FH testing enhances the benefit to other family members, as the autosomal dominant pattern ensures that 50% of members will be at risk.

Conclusions

- School based programs are the most effective and equitable means of identifying FH and should be supported in a collaborative manner with health care systems, industry, and the community.
- With availability to all students, diversity, equity, and inclusion concerns are addressed and provide at risk populations with personalized health information.
- The results from this study suggest school-based screening programs are the best mechanism for FH screening.

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