

Development of an application to support the identification of patients with familial hypercholesterolaemia

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Background: Familial hypercholesterolaemia (FH) is associated with the long-term elevation of cholesterol levels in the blood. According to guidance from the National Institute for Health and Care Excellence (NICE), FH is suspected if total cholesterol exceeds 7.5 and 9 mmol/L in people under and over 30 years of age, respectively. The use of these cut-offs may over diagnose older people whose cholesterol has risen due to lifestyle factors, and underdiagnose younger people who have not reached the threshold, but may be at risk.

Aims: To develop an interactive application which places a patient on a specific population-based cholesterol centile according to their age and sex to improve the identification of people at risk of FH.

Methods: Health Survey for England (HSE) data were obtained from NHS Digital covering seven years between 2003 and 2014. Data for age, sex, high-density lipoprotein cholesterol (HDL-C), total cholesterol (TC), and use of lipid-lowering drugs were extracted. Centiles were derived at intervals of 0.1 between 0.5 and 99.5, for non-HDL cholesterol (non-HDL-C) [non-HDL-C = TC – HDL-C] and TC, in patients not being treated with lipid-lowering drugs.

Results: An interactive application was developed using Shiny that places a patient on a specific cholesterol centile based on their age and sex. Figure 1 shows an example of a 35 (I) and 55 (II) year old male with a TC of 9 mmol/L, which places the example patients on the 98.7 and 96.9 centiles, respectively.

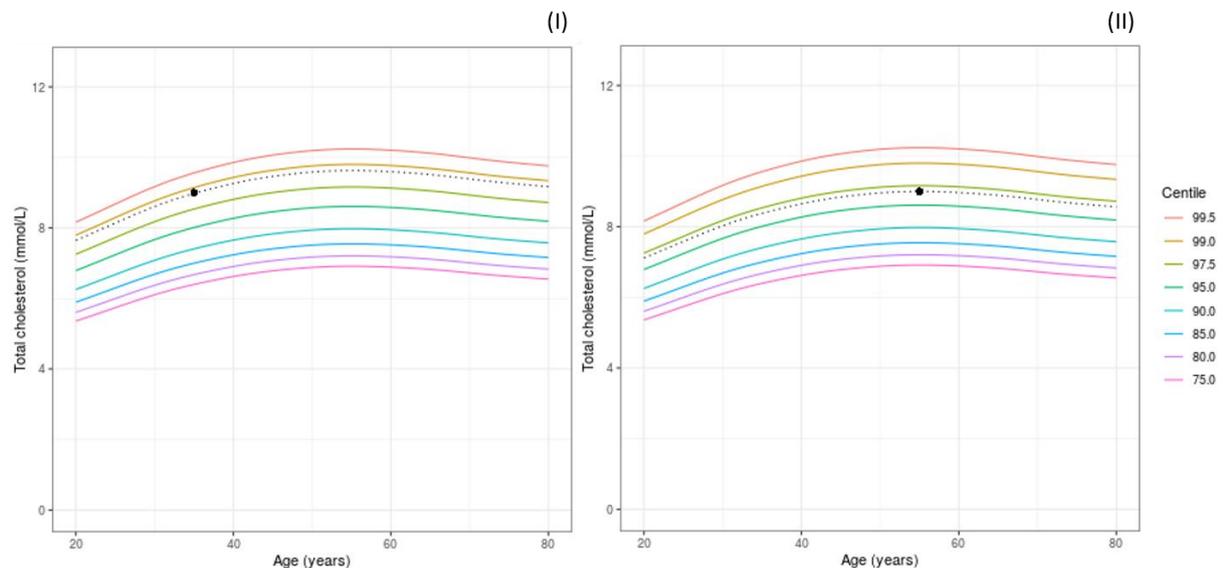


Figure 1 – Screenshots from the interactive Shiny application (<https://micncltools.shinyapps.io/miccentilesshinyapp/>) showing total cholesterol centile plots for a 35 (I) and 55 (II) year old male with a total cholesterol of 9 mmol/L. The two example patients are each indicated by a black dot on the plots

Conclusions: When used in conjunction with current methods, the use of age and sex adjusted cholesterol centiles could help improve the identification of patients with FH, and therefore refine the selection of index cases for targeted genetic testing.

Keywords

Familial hypercholesterolaemia, cholesterol, identification, application.