Letter by Paton Regarding Article, “Knowing the Prevalence of Familial Hypercholesterolemia Matters”

To the Editor:
I read with great interest the editorial1 regarding the prevalence of familial hypercholesterolemia (FH) in the United States and why the early recognition of this condition is so essential for the optimal management of individuals with the condition, in particular, because the PCSK9 monoclonal antibodies, alirocumab and evolocumab, were approved by the US Food and Drug Administration in 2015 for the treatment of heterozygous FH. However, although evolocumab was also approved for use in homozygous FH, optimal drug therapy of patients with homozygous FH still left their low-density lipoprotein cholesterol levels well above 4.9 mmol/L.2

An essential part of the diagnosis of FH1 was a low-density lipoprotein cholesterol of ≥4.9 mmol/L (≥190 mg/dL). However, patients with cholesteryl ester storage disease, also known as lysosomal acid lipase deficiency, may also have a low-density lipoprotein cholesterol of ≥4.9 mmol/L.3 Such patients are very likely to have also hepatomegaly and raised levels of alanine aminotransferase but do not have an autosomal dominant inheritance pattern, cholesteryl ester storage disease being an autosomal recessive disorder. A diagnostic algorithm has been proposed3 that includes consideration of these and other aspects.

The levels of lysosomal lipase activity can be measured fluorometrically in dried blood spots to confirm the diagnosis of cholesteryl ester storage disease.4 It is important to distinguish patients with cholesteryl ester storage disease from those with FH, because they can now be successfully treated using sebelipase alfa to replace the enzyme they lack.5

DISCLOSURES
None.

AFFILIATION
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REFERENCES

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