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LETTER TO THE EDITORS

Rebuttal to "ezetimibe treatment should be considered for patients with sitosterolemia"

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We have presented an adolescent of Amish descent with atherosclerotic renal artery stenosis as a cause for hypertension. He was subsequently diagnosed with sitosterolemia, a rare autosomal recessive genetic disorder characterized by the retention of both plant sterols and cholesterol in affected individuals. Determination of his serum sterol profile by gas chromatography revealed 91.2 % cholesterol (normal >99 %), with elevated plant sterol levels (normal <1 %) at 2.7 % campesterol, 5.6 % beta-sitosterol and 0.5 % stigmasterol; these findings were consistent with sitosterolemia [1]. His genetic testing has not yet been completed, but the clinical picture of the patient and elevated serum plant sterols support the diagnosis of sitosterolemia. The patient was initially placed on atorvastatin. After he was diagnosed with sitosterolemia he was started on ezetimibe and resin colestid. We agree with Hu et al. that ezetimibe is the drug of choice for sitosterolemia [2]. Ezetimibe binds directly to Niemen Pick C1-like 1 (NPC1L1) and inhibits absorption of sitosterols from the intestinal epithelium. In a multicenter study 37 patients were randomized to receive placebo ($n=10$) versus ezetimibe ($n=30$) for 8 weeks. Ezetimibe treatment resulted in a significant reduction in sitosterol and campesterol levels [3].

We believe a multidrug approach is warranted to treat sitosterolemia in order to prevent premature atherosclerotic cardiovascular disease. Although there is no doubt that ezetimibe is beneficial for patients with sitosterolemia, close monitoring for future side effects is warranted particularly in children, considering their longer life span.

References

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