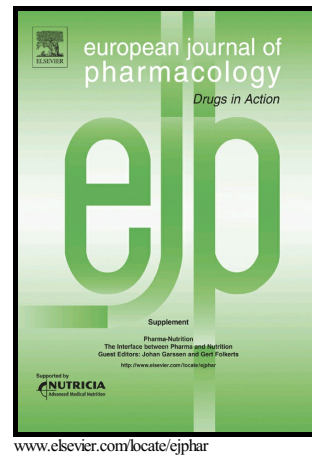


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Monogenetic disorders of the cholesterol metabolism and premature cardiovascular disease

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Abstract

Cholesterol is of vital importance for normal function of organisms. However, a high serum level is associated with an increased risk of cardiovascular disease. In this review an overview is presented of the different known monogenetic disorders of the cholesterol metabolism which lead to unfavourable lipid profiles from childhood onwards and premature cardiovascular disease. Since these monogenetic disorders have a large variety in clinical presentation, ranging from scarcely any to extreme premature cardiovascular disease, the frequency is underestimated and often the correct diagnosis is not made. This results into a missed opportunity for optimal treatment as well as for appropriate counselling of family members. Therefore, greater awareness among physicians is needed.

Keywords

Premature cardiovascular disease, cholesterol metabolism, monogenetic disorders; Familial hypercholesterolemia, LDL-C, HDL-C

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