- The true prevalence of homozygous and heterozygous familial hypercholesterolemia (HoFH and HeFH, respectively) is currently underestimated, with worldwide variation in its diagnosis.
- Untreated patients with HoFH usually have significant morbidity and mortality by the second decade of their lives.
- Diagnosis requires a combination of clinical manifestation of the disease, family history, lipid levels, and genetic testing .
- HeFH and HoFH should be managed using a combination of statins, ezetimibe, bile acid sequestrants, and lipoprotein apheresis.
- Newer agents such as lomitapide, mipomersen, and pro-protein convertase subtilisin/kexin 9 (PCSK9) inhibitors are providing vital therapeutic alternatives.

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