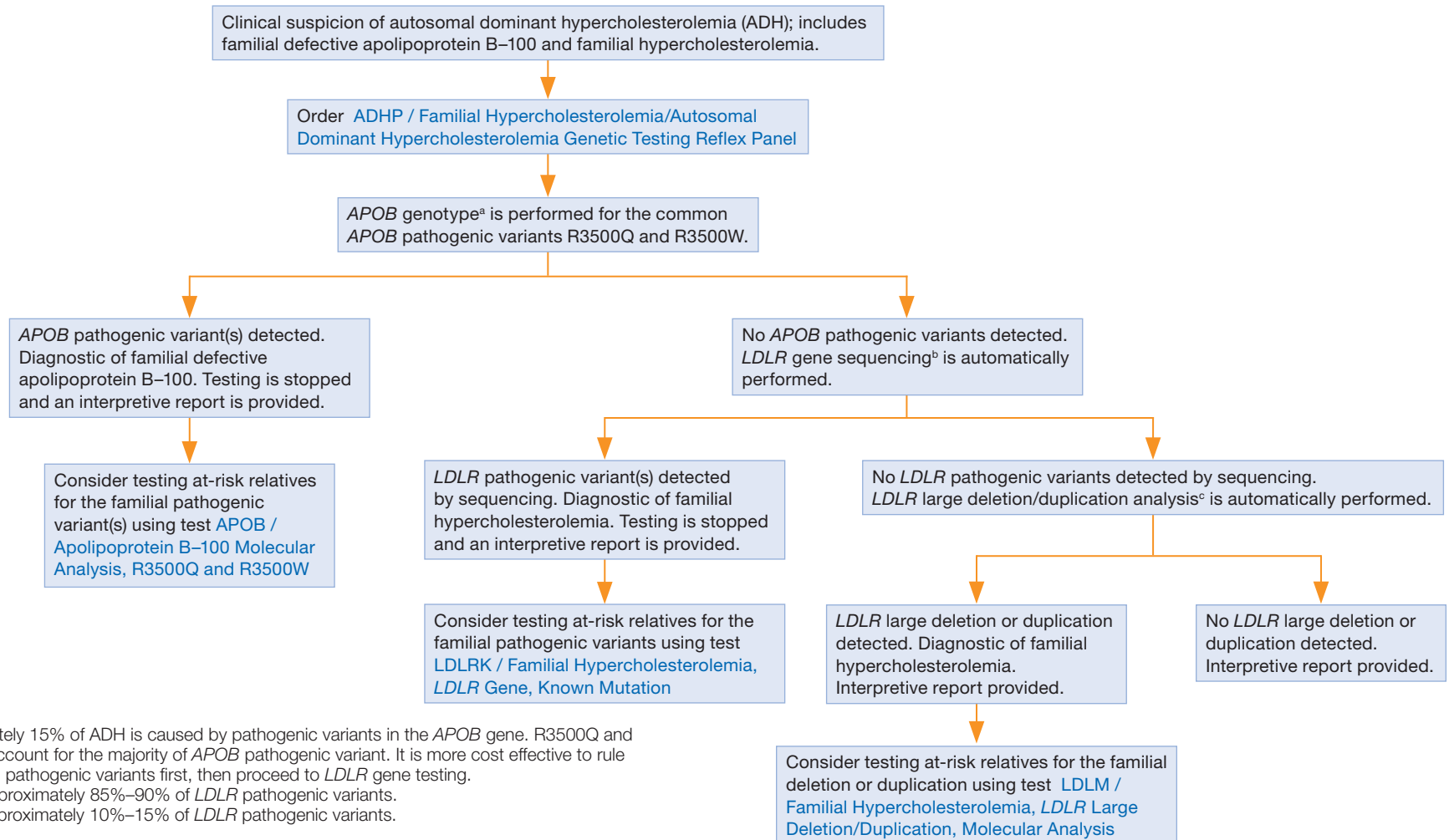


Familial/Autosomal Dominant Hypercholesterolemia Diagnostic Algorithm



^a Approximately 15% of ADH is caused by pathogenic variants in the *APOB* gene. R3500Q and R3500W account for the majority of *APOB* pathogenic variant. It is more cost effective to rule out these 2 pathogenic variants first, then proceed to *LDLR* gene testing.

^b Detects approximately 85%–90% of *LDLR* pathogenic variants.

^c Detects approximately 10%–15% of *LDLR* pathogenic variants.

Note: If homozygous or compound heterozygous familial hypercholesterolemia is suspected based on clinical presentation, additional testing for a second pathogenic variant may be appropriate. The laboratory will contact the client and/or provider to discuss adjustment of this algorithm in appropriate cases.