Amyloidosis (Familial) Test Algorithm*

Clinical findings suggestive of familial amyloidosis

Order TTRX / Amyloidosis, Transthyretin-Associated Familial, Reflex, Blood
- Test begins with transthyretin (TTR) protein analysis by mass spectrometry (MS)
- DNA sequencing is automatically performed when appropriate

Abnormal TTR protein variant observed by MS
- ATTRZ / TTR Gene, Full Gene Analysis, Varies
- Disease-causing mutation identified—diagnostic for TTR-associated familial amyloidosis
- Variant of uncertain significance (VUS) identified
- Nondisease-causing polymorphism identified
- No alteration identified

If clinical suspicion of TTR-associated amyloidosis remains high, or there is a positive family history, order DNA sequencing (ATTRZ / TTR Gene, Full Gene Analysis, Varies)

Possible explanations:
- Normal TTR protein
- Abnormal TTR protein with undetectable change in mass (~10% of patients)

No TTR protein variant observed by MS

No alteration identified
- Does not rule out other causes of familial amyloidosis
- Consider alternative diagnosis

Diagnosis of amyloidosis**

*Interpretive report provided for all tests in this algorithm.
**See Laboratory Approach to the Diagnosis of Amyloidosis algorithm.