Amyloidosis (Familial) Test Algorithm*

**Diagnosis of amyloidosis**

- 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**

- Disease-causing mutation identified—diagnostic for TTR-associated familial amyloidosis

- Variant of uncertain significance (VUS) identified

- Nondisease-causing polymorphism identified

- No alteration identified

- Does not rule out other causes of familial amyloidosis
- Consider alternative diagnosis

**No TTR protein variant observed by MS**

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

*Interpretive report provided for all tests in this algorithm.

**See “Laboratory Approach to the Diagnosis of Amyloidosis” algorithm.**