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

No TTR protein variant observed by MS

ATTRZ / *TTR* Gene, Full Gene Analysis

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).

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

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

Variant of uncertain significance (VUS) identified

Nondisease-causing polymorphism identified

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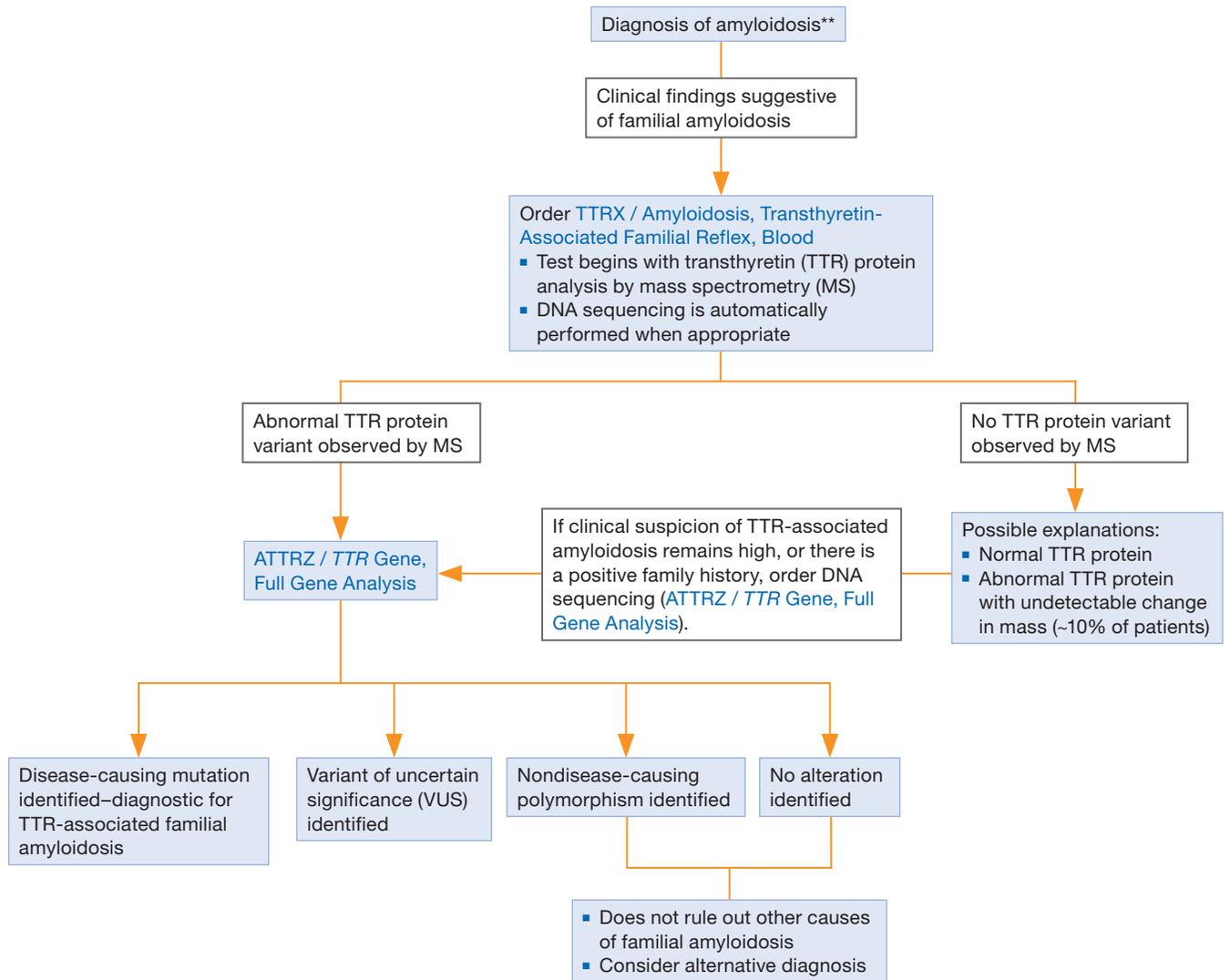
- Does not rule out other causes of familial amyloidosis
- Consider alternative diagnosis

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

08/2015