

Using Genetic Testing for Diagnosis and Risk Assessment for Transthyretin Amyloidosis (ATTR)

Indications:

- Clinical diagnosis of ATTR
- Congestive heart failure, often restrictive, and often accompanied by carpal tunnel syndrome
- Known familial *TTR* gene mutation
- Biochemically-proven transthyretin amyloid protein deposition in biopsy specimens

Benefits:

Genetic testing for ATTR

- Can confirm a clinical diagnosis of ATTR and identify the familial mutation(s)
- Can differentiate between systemic and cardiac-specific ATTR, depending upon the exact mutation found
- Can, in some cases, inform the prognosis based on the nature of the familial mutation
- Can identify carriers of known familial mutations in relatives of patients, alerting them to their increased susceptibility to ATTR
- Can identify candidates for liver and/or heart transplantation at early stages of disease

Background:

- ATTR is an autosomal dominant disease generally characterized by progressive neuropathy, cardiomyopathy, nephropathy, rheumatopathy, CNS abnormalities, and/or vitreous opacities, which can manifest individually or in combinations.^{1,2}
- ATTR affects approximately 1 out of every 100,000 persons worldwide.^{1,2}
- The cardiac-specific form of ATTR is characterized by restrictive cardiomyopathy, coronary insufficiency, valvular thickening, conduction system disease, and arrhythmias.³
- Patients with the cardiac-specific ATTR typically present with symptoms of congestive heart failure between the ages of 60 and 70 years and may also present with carpal tunnel syndrome.^{3,4}
- Since cardiac-specific ATTR has been associated with mutations in *TTR*, genetic testing can confirm a diagnosis of ATTR as the cause of congestive heart failure.²
- Liver transplantation is the only effective treatment since the source of transthyretin production is removed. In cardiac-specific ATTR, it is recommended that both the heart and liver are transplanted at early stages of disease.^{1,2,3}

References: 1. Benson MD, et al (2009) *Genetics: Clinical Implications of TTR Amyloidosis*. In *Recent Advances in Transthyretin Evolution, Structure and Biological Functions*: Springer Berlin Heidelberg. 173. 2. Benson MD (2003) *Best Pract Res Clin Rheumatol* 17:909. 3. Falk RH, et al (2007). *Prog Cardiovasc Dis* 52:347. 4. Gavin A, et al (2007) *J R Coll Physicians Edinb* 38: 196.

Ordering Information: Please see other side.



Ordering Information for Transthyretin Amyloidosis (ATTR) Testing

Indications for Testing

- Clinical diagnosis of ATTR
- Congestive heart failure, often restrictive, and often accompanied by carpal tunnel syndrome
- Known familial *TTR* gene mutation
- Biochemically-proven transthyretin amyloid protein deposition in biopsy specimens

Ordering Information

Gene(s)	Test Code
<i>TTR</i>	252810

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.