



# Screening Tool to Recognize Amyloidosis

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# Disclosures

**Consulting Fees/Honoraria:** Pfizer, Akcea/Ionis, Alnylam, Takeda, Sanofi-Genzyme, Astra-Zeneca, Bayer

**Clinical Trials:** Pfizer, Eidos, Akcea/Ionis, Alnylam

**Speaker Fees:** Pfizer, Akcea/Ionis, Alnylam,

**Research Grants:** Pfizer, Akcea/Ionis, Alnylam, Eidos

**Educational Grants:**





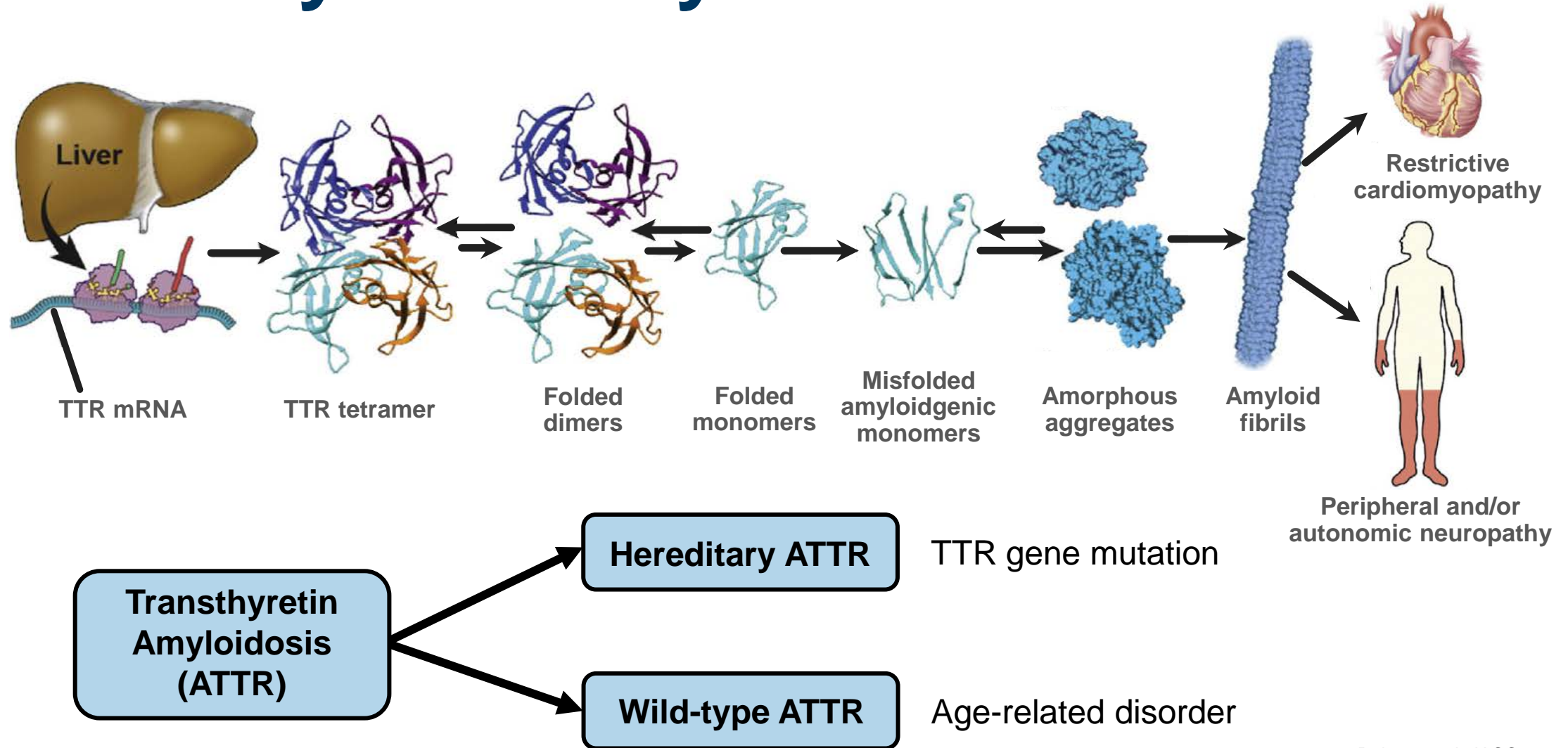
# Learning Objective


- Discuss patient questionnaires that can be used by cardiologists to help screen for signs and symptoms of amyloidosis to facilitate earlier disease recognition and diagnosis





# Transthyretin Amyloidosis - ATTR





**Cardiac amyloidosis suspected** based on standard heart failure workup, including cardiac imaging with either echocardiography and/or CMR, troponin and BNP/NT-proBNP

**Screen for plasma cell dyscrasia** – serum and urine protein electrophoresis with immunofixation, serum free light chain assay

**AL amyloidosis suspected** – monoclonal protein present

**Hematology referral** – biopsy of involved organ, typically EMB, renal, BMB or fat pad (which cannot exclude systemic amyloidosis) with MS or IHC if positive

**AL cardiac amyloidosis** – (or other type by EMB with MS or IHC)

Cardiac amyloidosis excluded

**ATTR amyloidosis suspected** – monoclonal protein absent

**Tc-99m-PYP scan** – if unavailable, perform EMB with MS or IHC if positive

**ATTR cardiac amyloidosis** – perform TTR genetic testing

Positive – **hATTR**

Negative – **wtATTR**

Cardiac amyloidosis excluded – if equivocal results, consider EMB



# ATTR – Hiding in Plain Sight

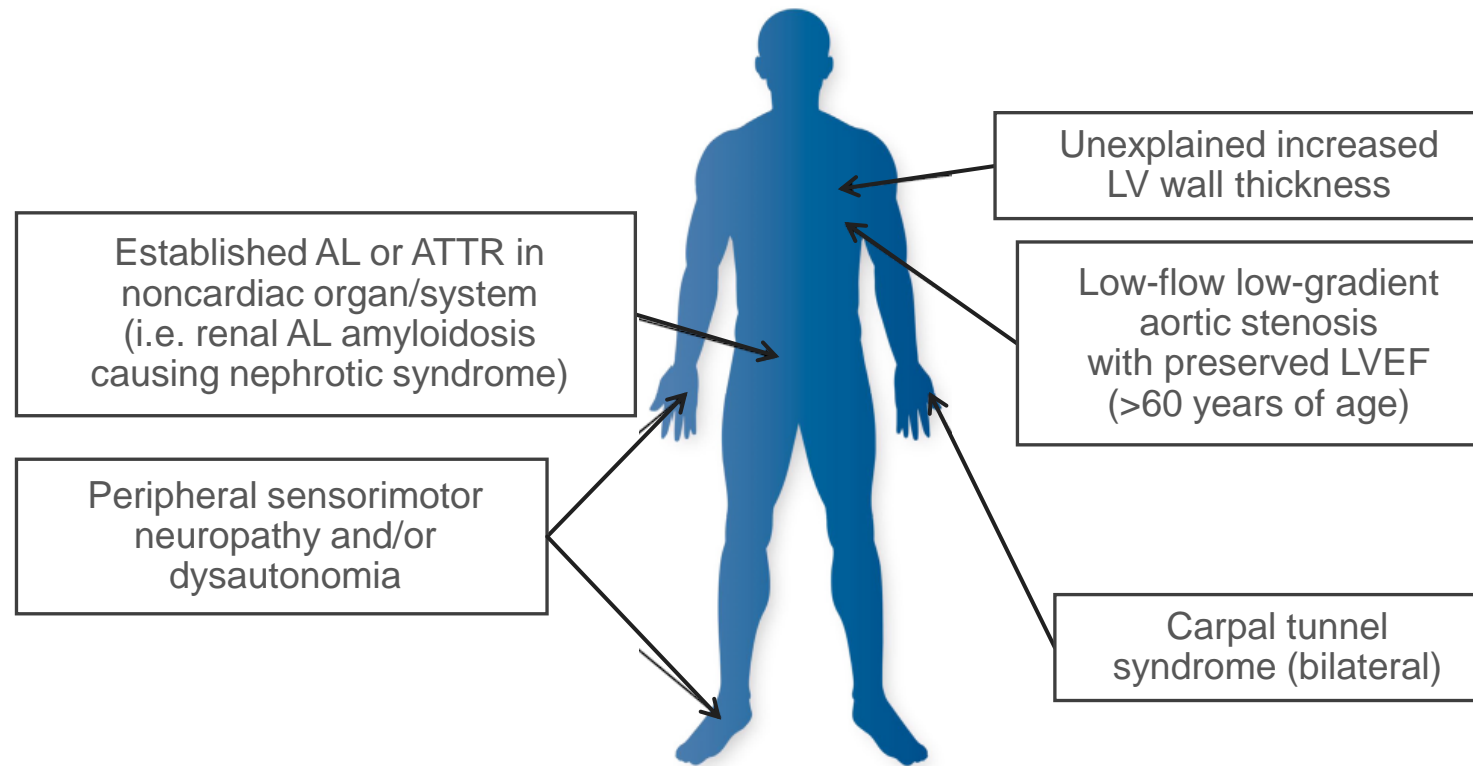
- Nonspecific signs, symptoms, and findings on diagnostic work-up
  - Wide range of clinical presentations
  - Multiple types of tests available
- Very challenging to recognize in busy clinical practice, especially in early stages
- **Awareness** and **index of suspicion** are critical for early disease recognition to facilitate treatment initiation and attenuation of disease progression





# Diagnostic Workup for Cardiac Amyloidosis

SUSPECT CARDIAC AMYLOIDOSIS WHEN NEW  
ONSET HEART FAILURE WITH  $\geq 1$  OF THE FOLLOWING





# ATTR Patient Questionnaires

- Initially developed to assist specialists with recognition of ATTR manifestations outside of their specialty area
  - Created by the Amyloidosis Program of Calgary
- Short patient questionnaires focused on type and burden of symptoms, medical history and family history
- Can be completed on-paper or electronically
- Different versions that can be used in different clinical settings
  - Cardiomyopathy
  - Neuropathy
  - General (multi-system)
  - Comprehensive



# ATTR Patient Questionnaires

## Transthyretin Cardiomyopathy

### Patient Questionnaire (Standard)

[Clear All](#)[Print](#)[Save](#)[Email](#)

Patient Name \_\_\_\_\_

#### Symptoms:

- 1 Do you feel full/bloated easily after meals?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 2 Do you feel excessively tired/lethargic?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 3 Do you experience abdominal (stomach) pain?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 4 Do you experience constipation?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 5 Do you experience diarrhea/loose or watery bowel move  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 6 Do you experience blurred vision (even with corrective eye-v  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 7 Do you experience sexual dysfunction?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 8 Have you experienced unintentional weight loss?  
☐ No ☐ Yes
- 9 Do you require an aid to walk and/or move around?  
☐ No ☐ Cane ☐ Walker ☐ Wheelchair

1 of 2

## Transthyretin Amyloidosis

### Cardiomyopathy Questionnaire

[Clear All](#)[Print](#)[Save](#)[Email](#)

Patient Name \_\_\_\_\_ Date \_\_\_\_/\_\_\_\_/\_\_\_\_  
(mm/dd/yyyy)

#### Symptoms:

- 1 Do you suffer from shortness of breath?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 2 If yes, what level of activity makes you short of breath?  
☐ Two flights of stairs (20 steps) ☐ One flight of stairs (10 steps) ☐ Walking around your home  
☐ At rest ☐ Not applicable
- 3 Do you have swelling (fluid retention) in your legs or other areas (waist, hands)?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 4 Do you need to prop your head up to breathe comfortably for sleeping?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 5 If yes, how many pillows do you use (how high do you prop your head-up)?  
☐ 2 pillows ☐ 3 pillows ☐ 4 pillows ☐ Sleep fully upright (i.e., in a chair)  
☐ Not applicable
- 6 Do you wake up in the middle of the night unable to breathe?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 7 Do you experience chest pain?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 8 Do you feel full/bloated easily after meals?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always

Continued »

1 of 2

## Transthyretin Amyloidosis

### Patient Questionnaire

[Clear All](#)[Print](#)[Save](#)[Email](#)

Patient Name \_\_\_\_\_

#### Symptoms:

- 1 Do you suffer from shortness of breath?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 2 If yes, what level of activity makes you short of breath?  
☐ Two flights of stairs (20 steps) ☐ One flight of stairs (10 steps)  
☐ At rest ☐ Not applicable
- 3 Do you have swelling (fluid retention) in your legs or other  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 4 Do you need to prop your head up to breathe comfortably  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 5 If yes, how many pillows do you use (how high do you prop  
☐ 2 pillows ☐ 3 pillows ☐ 4 pillows ☐ Sleep fully upright  
☐ Not applicable
- 6 Do you wake up in the middle of the night unable to breathe  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 7 Do you experience chest pain?  
☐ Never ☐ Some of the time ☐ Most of the time ☐
- 8 Do you feel full/bloated easily after meals?  
☐ Never ☐ Some of the time ☐ Most of the time ☐

1 of 4

## Transthyretin Amyloidosis

### Neuropathy Questionnaire

[Clear All](#)[Print](#)[Save](#)[Email](#)

Patient Name \_\_\_\_\_ Date \_\_\_\_/\_\_\_\_/\_\_\_\_  
(mm/dd/yyyy)

#### Symptoms:

- 1 Are you unable to differentiate hot from cold (for example, when getting in the shower or bath)?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 2 Are you unable to sweat even when you are hot?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 3 Do you feel numbness, tingling, burning or prickling sensation in the hands or feet?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 4 Do you have difficulty with balance (for example, in the shower or at night time or other times)?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always
- 5 Do you require an aid to walk and/or move around?  
☐ No ☐ Cane ☐ Walker ☐ Wheelchair
- 6 Do your hands or arms ever "fall asleep", go "dead" or get numb during the night?  
☐ Never ☐ Some of the time ☐ Most of the time ☐ Always

Continued »

1 of 2



# ATTR Patient Questionnaires

- Not validated
- Not a risk score
- Do not incorporate physical examination or investigation findings
- Educational and clinical tool



# ATTR Patient Questionnaires

- Thank you
- Questions / comments?
  - We want your feedback!
- [nmfine@ucalgary.ca](mailto:nmfine@ucalgary.ca)