HEREDITARY ATTR AMYLOIDOSIS OVERVIEW

Amyloidosis Research Consortium

WHAT IS HEREDITARY ATTR (hATTR) AMYLOIDOSIS?

Hereditary ATTR amyloidosis is caused by a mutation or change in the transthyretin (TTR) gene which is inherited (i.e. runs in families). The mutation results in an abnormal TTR protein that is unstable and misfolds, forming amyloid deposits in various organs of the body.

WHAT ARE SOME SYMPTOMS OF hATTR AMYLOIDOSIS?

While not a complete list, some of the more common symptoms associated with hATTR are:

Heart Symptoms

Abnormal heart rhythms, shortness of breath, leg/feet swelling, and/or fainting.

Nervous System Symptoms

Tingling in the hands or feet, numbness in hands or feet, weakness in your extremities, burning in your extremities, and/or loss of movement control

Other Symptoms

- Gastrointestinal (GI) symptoms including diarrhea, nausea and/or digestive issues
- Frequent urinary tract infections
- Symptoms related to muscles, tendons and joints including carpel tunnel syndrome, lumbar spine stenosis, or bicep tendon rupture

HOW IS hATTR DIAGNOSED?

Diagnosing hATTR requires genetic testing to identify the TTR gene mutation. Other tests may include an Electrocardiogram (EKG), Echocardiogram (ultrasound of your heart), Cardiac MRI and/or Technetium Pyrophosphate scan to confirm if there are abnormal proteins in your heart. Technetium Pyrophosphate scan is the most commonly used diagnostic test. A heart biopsy may also be considered. These tests would be done by seeing a cardiologist or heart doctor.

To confirm the diagnosis of hATTR in your nervous system you may need a Nerve Conduction Study (NCS) or Electromyography (EMG). This would be done by seeing a neurologist or nerve doctor.

320 NEVADA STREET, NEWTON, MA 02460

HEREDITARY ATTR AMYLOIDOSIS

Amyloidosis Research Consortium

IMPORTANT NOTE

Genetic testing is required to diagnose hereditary type amyloidosis. This may tell you if and which pathologic gene could be causing hATTR.

You can be a **carrier** of this gene and **not develop** the condition or "disease state" of hATTR.

TREATMENT AND MANAGEMENT

There were no treatment options prior to 2019 and we now have multiple FDA approved treatments (some for cardiomyopathy or enlarged heart and others for peripheral neuropathy). There are many other treatments in development. You may need other medications to help with symptoms such as diuretics (water pills) or nerve pain medications.

PREVENTION STRATEGIES

If you have any of the symptoms mentioned, please do not be afraid to talk with your healthcare team. Be your own advocate. If the disease is suspected, get yourself and family members tested. If you carry the gene, be aware of symptoms so you can discuss with your healthcare team and get early treatment. Early diagnosis and treatment can help maintain your quality of life. The sooner the diagnosis, the better your chances improve for survival and quality of life.

ARC gratefully acknowledges Jennifer Fox, NP from Weill-Cornell NYP for her contributions to the development of this document.

Ask ARC Your Questions:
(617) 467-5170 or support@arci.org

320 NEVADA STREET, NEWTON, MA 02460