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ATTR Amyloidosis: Familial Vs Wild-Type

Announcer:
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Dr. Dasgupta:
This is CME on ReachMD, and I'm Dr. Noel Dasgupta. Today, I'm going to talk about the difference between hereditary, or familial, transthyretin amyloidosis versus wild-type, or age-related, amyloidosis.

So amyloidosis is a protein-folding disorder. The protein, transthyretin, may misfold and deposit in organs and tissues. This misfolding occurs because of either genetic changes or something you may inherit from your parents due to changes in your DNA or are related to aging. The age-related form is called wild-type amyloidosis because the gene is in its wild, or native, form.

Transthyretin is a protein that is normally produced by the liver and transports thyroid hormone and vitamin A. In transthyretin amyloidosis, the protein may misfold and deposit in organs and tissues.

The most common manifestations include cardiomyopathy which may present with heart failure with preserved ejection fraction early in the disease but reduced ejection fraction later in the disease, unexplained left ventricular hypertrophy, arrhythmias, and conduction disease. Some patients may also have valvular disease. Patients may also have polyneuropathy or nerve findings. This typically presents as numbness and tingling in the hands and feet. Later in the disease, patients may experience muscular weakness and may even become wheelchair bound. Autonomic dysfunction affecting the ability to stand and gastrointestinal functions may also be affected. Soft tissue manifestations, including carpal tunnel syndrome and spinal stenosis, are often associated with this disease.

It is important to determine if it is a hereditary or age-related form of amyloidosis because of the familial implications. Transthyretin amyloidosis of the hereditary type is an autosomal-dominant disease. This means that half of offspring may have the gene that puts them at risk for developing transthyretin amyloidosis. Although they may inherit the gene, they may not develop the disease. Genetic testing can be performed by either doing a blood sample or a saliva sample.

In general, transthyretin amyloidosis is a disease of older patients. Most commonly, it affects individuals over age 60, but in rare cases, it can occur earlier. In the familial or hereditary form, symptoms may occur up to 10 years or more earlier.

Treatment for transthyretin amyloidosis focuses on either decreasing the amount of production of the transthyretin protein which is made by the liver, or stabilizing the transthyretin protein after it is made by the liver so that it does not misfold and deposit in organs and tissues.

Early diagnosis and treatment is important to help prevent end-organ dysfunction. The earlier treatment is started, the less likely the patient will have severe organ damage.

The therapies for transthyretin amyloidosis typically stabilize the patients and, in some cases, improve the patient's symptoms. Therefore, it's important to start treatment early.

Genetic testing is important to differentiate between hereditary and wild-type amyloidosis. This is important because hereditary disease may affect other family members. In the United States, 3% to 4% of African Americans have a genetic change in the transthyretin gene that puts them at risk for hereditary transthyretin amyloidosis.

Transthyretin amyloidosis is a disease that is much more common than we previously thought. It is a multisystem disease that may affect both the heart, nervous system, and soft tissues, as well as other organs. A high clinical suspicion is necessary to try to integrate findings and make a diagnosis. This is important because if it's a hereditary form, other family members are at risk.

Well, that was brief, but thank you for listening. This is Dr. Noel Dasgupta and ReachMD CME.

Announcer:
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