Dear Family Member:

This letter is to notify you that our family has been diagnosed with a genetic condition known as hereditary transthyretin (hATTR) amyloidosis. This condition is also known as familial amyloidosis, hereditary amyloidosis, ATTRmt, ATTRm, FAP and FAC. Disease symptoms result from an abnormal buildup of a protein, called amyloid, in the nerves (neuropathy), heart (cardiomyopathy), and gastrointestinal (GI) tract. The purpose of this letter is to familiarize you with hATTR amyloidosis, implications for family members, and the steps that you can take to learn more about your risk.

The symptoms of hATTR amyloidosis can vary widely among people with the same mutation and even within families. Symptoms usually develop in adulthood, although the age at which initial symptoms appear can vary. Symptoms may include shortness of breath, fatigue, numbness or tingling in the hands and feet, carpal tunnel syndrome, and constipation and/or diarrhea. Of importance, not everyone with a mutation will develop symptoms in their lifetime. **Identifying those who are at risk for developing hATTR amyloidosis may allow for earlier and more effective symptom detection and management.**

hATTR amyloidosis is due to gene alterations (known as mutations) in the TTR gene. Through genetic testing, it was found that I carry an alteration in the TTR gene (<___>), which is the cause of hATTR amyloidosis in our family. My genetic testing was performed at <___>.

First degree-relatives (parents, siblings, children) are at 50% risk to also carry this TTR gene alteration. Now that the specific gene change has been identified in our family, genetic testing can identify who else in our family is at risk for developing hATTR amyloidosis. Relatives who carry the gene alteration can take steps to manage symptoms or potentially reduce risk of developing symptoms. Relatives who do not carry the gene alteration are not at increased risk for developing hATTR amyloidosis or passing it on to their children.

It is important that you discuss this with a physician or genetic counselor who can provide more information about screening recommendations and the option of genetic testing. Dr. _____ and the genetic counselors at the _____ can provide you with more information and help with genetic testing. To schedule an appointment, please call _____. Alternatively, a genetic counselor near you can be located by visiting the National Society of Genetic Counselors website (http://nsgc.org) and clicking on the button called “Find a Genetic Counselor.”

While it can be concerning to learn that there is a risk for a genetic condition in the family, there are many resources available for families at risk for hATTR amyloidosis, and I want to help ensure that our family has access to this important information.

Sincerely,