Primary hyperoxaluria is a rare inherited disease caused by a gene change (mutation) that causes calcium oxalate to build up faster than the kidneys can excrete it. This results in recurrent kidney or bladder stones, often occurring at a very young age. Other parts of the body, including the nervous system, heart, retina, and skin, can also be damaged if excess calcium oxalate accumulates. There are three types of primary hyperoxaluria: Type 1, Type 2, and Type 3. Primary hyperoxaluria Type 1 (PH1) is the most common form and accounts for about 80% of all cases.

WHAT IS PRIMARY HYPEROXALURIA?

Primary hyperoxaluria is caused by a mutation in the AGXT gene. Genetic testing evaluates whether a person has a mutation in this gene. Testing can give several possible results: a positive result, a negative result, or an uncertain result.

We all have two copies of the AGXT gene, one inherited from each parent. When a test result is positive, it means that both copies of the AGXT gene have a mutation, and the person has an increased risk to develop symptoms of PH1 during his or her lifetime.

WHAT A POSITIVE GENETIC TEST FOR PRIMARY HYPEROXALURIA TYPE 1 MEANS

PH1 is caused by a mutation in the AGXT gene. Genetic testing evaluates whether a person has a mutation in this gene. Testing can give several possible results: a positive result, a negative result, or an uncertain result.

We all have two copies of the AGXT gene, one inherited from each parent. When a test result is positive, it means that both copies of the AGXT gene have a mutation, and the person has an increased risk to develop symptoms of PH1 during his or her lifetime.

WHAT A POSITIVE GENETIC TEST MEANS FOR YOUR FAMILY

A positive genetic test result may also have important health and social implications for your family members, as PH1 is passed down through families.

Typically, the parents of a person who has PH1 are not affected by the disease. Only one of their two copies of the AGXT gene has a mutation. The other copy works normally and is enough to prevent excess buildup of calcium oxalate. If the parents of a person who has PH1 have more children, each child has a 25% risk of being affected with PH1, and a 50% risk that one of their copies of the AGXT gene has a mutation.

When a person has a mutation in one of their copies of the AGXT gene, other family members may also have the same mutation. A conversation about PH1 may help you and your family members to plan future pregnancies. It can also help to increase understanding about living with PH1.

GENETIC COUNSELING

Genetic counseling may help you or your family members to better understand the risks, benefits, limitations, and potential implications of genetic testing. Genetic counseling should be performed by a trained health care professional and may be available before, during, or after genetic testing.

In addition to genetic testing, Alnylam is sponsoring no-charge third-party genetic counseling for individuals in the United States who may carry a gene mutation known to be associated with PH1.

To schedule telephone-based genetic counseling, call 1.888.475.3128 and reference the Alnylam Act® program.

Note: Callers will also be asked to provide a doctor’s name, address, phone, and fax number, as a detailed summary report will be sent directly to him/her following the genetic counseling session.