Physician Information

Unraveling USH:
An Usher Syndrome
Genetic Testing Initiative

Vivimus Enim WIVR Remedium

USHER SYNDROME COALITION

What is Usher syndrome?

Usher syndrome is an autosomal recessive condition that affects nearly 50,000 people in the United States. It is the most common genetic cause of combined deafness and blindness and is characterized by hearing loss or deafness, progressive vision loss and, in some cases, vestibular dysfunction. Over 95% of all currently known Usher-causing mutations lie in one of eight genes. The most sensitive genetic tests can now find the disease-causing mutations in more than 75% of Usher patients.

What is Unraveling USH?

Unraveling USH is a collaborative effort between the Usher Syndrome Coalition and the Stephen A. Wynn Institute for Vision Research at the University of Iowa (www.bit.ly/projectusher) to ensure everyone with Usher syndrome has access to genetic testing. Unraveling USH seeks to provide guidance and affordable testing to people who have been unable to get genetic testing in the past, either due to lack of access or financial barriers. This initiative will help to fulfill the Usher Syndrome Coalition's goal to identify everyone with Usher syndrome worldwide, while offering state-of-the-art genetic testing through the John and Marcia Carver Nonprofit Genetic Testing Laboratory at the University of Iowa.

Why is genetic testing important?

Genetic testing is the only way to get a definitive diagnosis of Usher syndrome. A genetic diagnosis will reveal the genetic subtype (USH1b, USH2a, USH3, etc.) of Usher syndrome. This information is often needed for participation in clinical trials, as many of the treatments in the research pipeline will depend upon knowing the exact genetic mutation and type of Usher one has. Genetic testing is vital to the identification of an adequate number of patients to qualify and enroll in clinical trials, paving the way for meaningful treatments.

A diagnosis through genetic testing can also give a better sense of the progression, leading to improved decisions about management and potential treatment.

How can physicians help their patients obtain genetic testing?

There are two paths for physicians:

1. If you choose to order the test for your patient, you will need to need to create an account through the Carver Laboratory and follow the instructions on the University of Iowa website (https://www.carverlab.org/request/physician).
2. You may choose to refer your patient to a specialty center or genetic counselor, who can coordinate testing. Genetic counseling resources can be found in the genetic testing Q&A section of the Usher Syndrome Coalition website (below).

Additional information can be found through the Usher Syndrome Coalition at www.usher-syndrome.org, under “Take Action.”