



Stephen A. Wynn
Institute for Vision Research

Dear *INSURANCE CARRIER*,

I am writing to ask that your company consider including the costs of genetic testing for Usher syndrome as a standard benefit for your customers.

Usher syndrome is an autosomal recessive eye condition that affects about 20,000 people in the United States. It is the most common cause of combined deafness and blindness. Over 95% of all currently known Usher-causing mutations lie in one of eight genes. The most sensitive tests can now find the disease-causing mutations in more than 75% of Usher patients.

Treatments for Usher syndrome can be divided into family planning, cochlear implants, hearing aids, gene therapy, stem cell therapy, and assistive devices. Newborn hearing testing coupled with genetic testing makes it possible to diagnose Usher syndrome much earlier than ever before. As a result, many of these treatment modalities are now more effective than they were in the past.

Couples who are carriers of the same Usher syndrome gene are at 25% risk (with each pregnancy) of having a child with Usher syndrome. Early diagnosis of the first child with associated genetic counseling makes it possible for a family to avoid having another affected child by various means ranging from adoption to pre-implantation genetic testing. The latter method involves in vitro fertilization and allows couples to have additional biological children while dramatically reducing the risk of having a second affected child.

Conventional hearing aids, provided at a very young age to a child with type 2 Usher syndrome, can dramatically improve the child's speech and educational development. These devices often allow children with profound hearing loss (as seen with type 1 Usher syndrome) to develop quite normal speech.

Usher patients with early stage disease may benefit from viral-mediated replacement of the dysfunctional gene. Such a treatment has already been devised for USH1B (MYO7A) and is now in human clinical trials.

Genetic testing by a philanthropically-supported non-profit laboratory such as the CLIA-certified Carver Laboratory at the University of Iowa is the most cost effective way to connect the small number of Usher patients covered by your company to the increasing number of valuable treatments available to them.

I urge you to consider genetic testing for Usher syndrome the "standard of care" for the individuals who have entrusted their health care to your company.

Sincerely,

A handwritten signature in blue ink, appearing to read "Edwin M. Stone".

Edwin M. Stone, M.D., Ph.D.
Director

University of Iowa Carver College of Medicine
375 Newton Road, 4111 MERF, Iowa City, Iowa 522452
319-335-8270 Fax 319-335-7142 edwin-stone@uiowa.edu