Gene Therapy 101

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*What is gene therapy?

*How does gene therapy work?

*Who is gene therapy for?

*When will gene therapy be available?
What is gene therapy?

A medical intervention designed to treat the genetic cause of a disease
Each one of our chromosomes contains many genes.

Most genes contain instructions for building functional proteins:

- **Normal gene**
  - **Normal protein**
  - **Normal function**
If there is a mistake in the instructions, the shape and function of the resulting protein are affected. This is the basis of genetic disease.

Gene containing an error

Improperly built protein

Abnormal function

Goal of gene therapy:
How does gene therapy work?

Therapies targeting a genetic error could take several forms:

1. Supply a normal copy of the instructions:

2. Read past the error:

3. Repair the error:

The therapeutic approach can vary depending on the gene and the type of error being treated.
Who is gene therapy for?

* People who have a genetic disorder for which the causative gene has been identified

VERY IMPORTANT TO HAVE A GENETIC DIAGNOSIS!

* People for whom the resumption of normal gene function will result in an improvement in their condition

STAGE OF DISEASE MUST BE KNOWN AND MONITORED
When will gene therapy be available?
When will gene therapy be available?

A clinical trial for a therapy to treat Usher syndrome type 1B is ongoing

Treatment availability in the future will depend on:
- progress in preclinical research
- success of clinical trials

This is a gene replacement therapy designed to treat vision loss in USH1B patients
Preclinical work is underway to develop the best gene therapy options for other forms of USH.

Waiting for this work to proceed is difficult, but there are a few things you can do:

- Know your genotype.
  www.usher-syndrome.org/take-action/get-genetic-testing.html

- Monitor your vision
  www.usher-registry.org/en/find_an_expert

- Stay connected via the USH Trust and Blue Book
  www.usher-registry.org
USH Talks

USHER SYNDROME COALITION