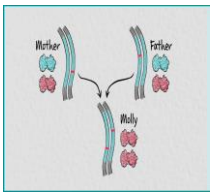

Genes as Medicine

hhmi | BioInteractive

Abbreviated Film Guide
Educator Materials

OVERVIEW

Gene therapy—the delivery of corrective genes into cells to treat a genetic disease—is an idea that was on scientists' minds as early as the 1960s. It took more than 35 years, however, to accumulate the knowledge and tools necessary to make gene therapy in humans a success. The HHMI short film *Genes as Medicine* tells the story of Drs. Jean Bennett, Albert Maguire, and their colleagues' decades-long effort to develop a gene therapy for a childhood disease called Leber congenital amaurosis (LCA).

KEY CONCEPTS

A. Some inherited diseases are caused by mutations in single genes. These mutations result in proteins that malfunction or, in some cases, no protein being produced, which cause the disease phenotypes.
B. For an individual to have a recessive genetic disease, they must have a disease-causing mutation in each copy (or allele) of a gene. If an individual has one allele with the mutation and one allele without it (in other words, they are heterozygous), they may have no disease symptoms.
C. Gene therapy is an experimental technique that adds corrective copies of mutated genes to a patient's cells.
D. Many biotechnology applications take advantage of naturally occurring processes. For example, in developing some gene therapies, scientists take advantage of viruses' ability to add genes to cells.
E. Most medical discoveries, including gene therapy, can take decades to move from the lab to new treatments available to people who need them.
F. Before most new medical treatments can be made available to patients, they must first be shown to work in animal models and then be shown to work in people taking part in clinical trials.

CURRICULUM CONNECTIONS

Standards	Curriculum Connections
NGSS (2013)	LS1.A, LS3.A, LS3.B
AP Biology (2015)	3.A.1, 3.C.1, 3.D.4, SP6
IB Biology (2016)	3.4, 3.5, B.4
Common Core (2010)	ELA.RST.9-12.2, WHST.9-12.4
Vision and Change (2009)	CC2, CC3

KEY REFERENCE

Russell, S., J. Bennett, J. A. Wellman, D. C. Chung, *et al*. (2017). Efficacy and safety of voretigene neparvovec (AAV2-hRPE65v2) in patients with *RPE65*-mediated inherited retinal dystrophy: a randomised, controlled, open-label, phase 3 trial. *The Lancet* 390:849-860.