

USH Talks

# Gene Therapy 101

Jennifer B. Phillips, Ph.D.

\*What is gene therapy?

\*How does gene therapy work?

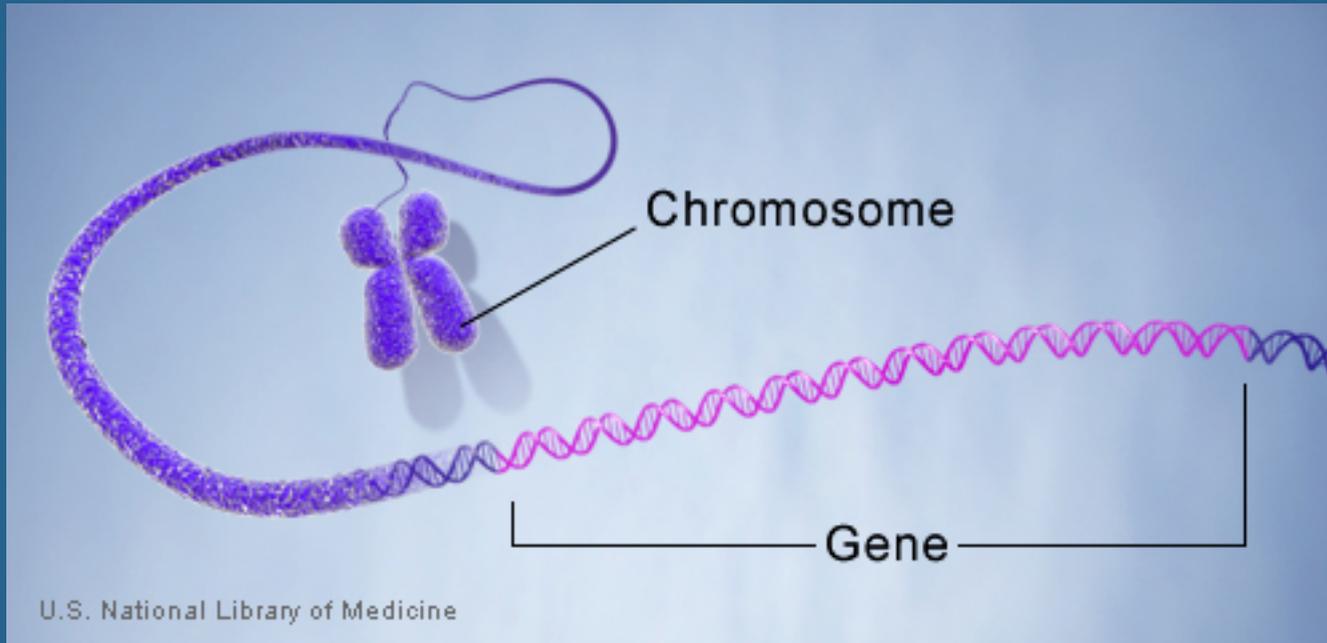
\*Who is gene therapy for?

\*When will gene therapy be available?

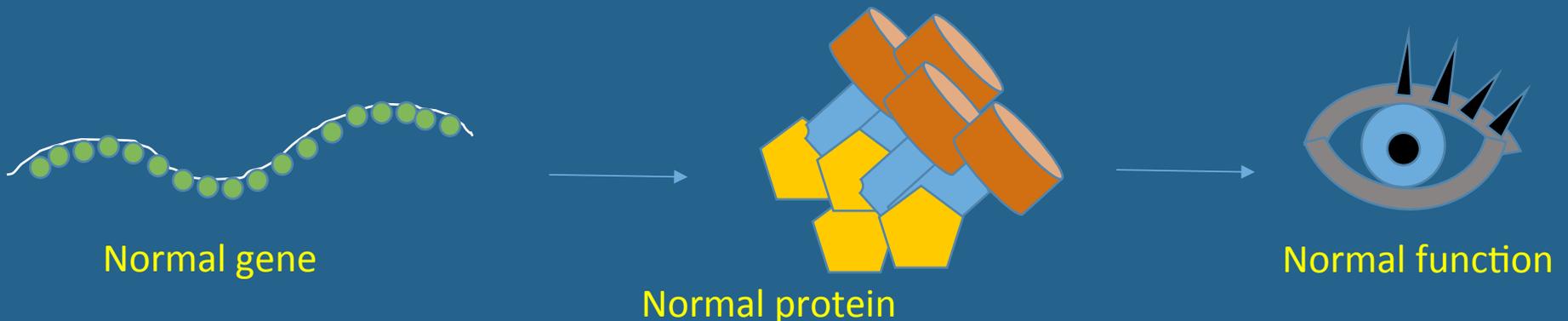
# What is gene therapy?

A medical intervention designed to treat the genetic cause of a disease

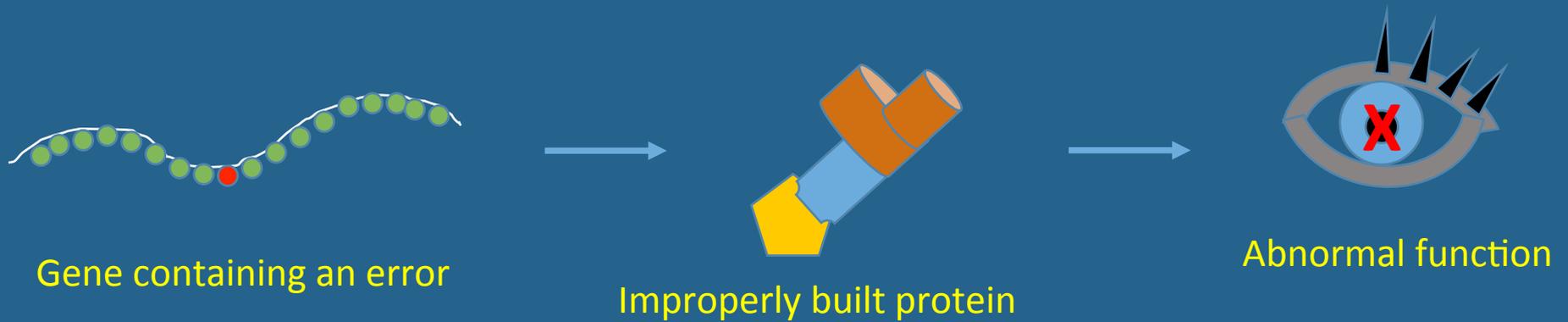
Each one of our chromosomes contains many genes



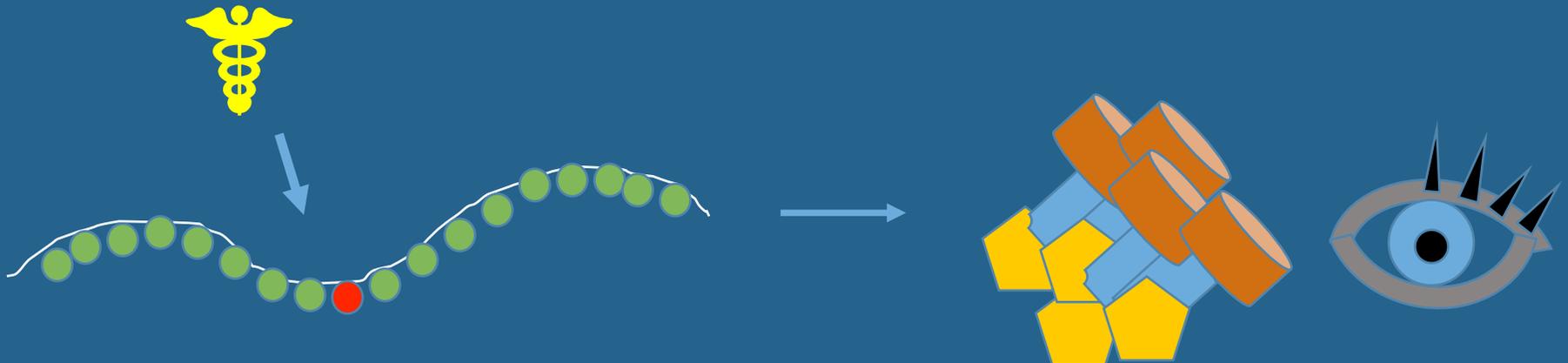
Most genes contain instructions for building functional proteins:



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.



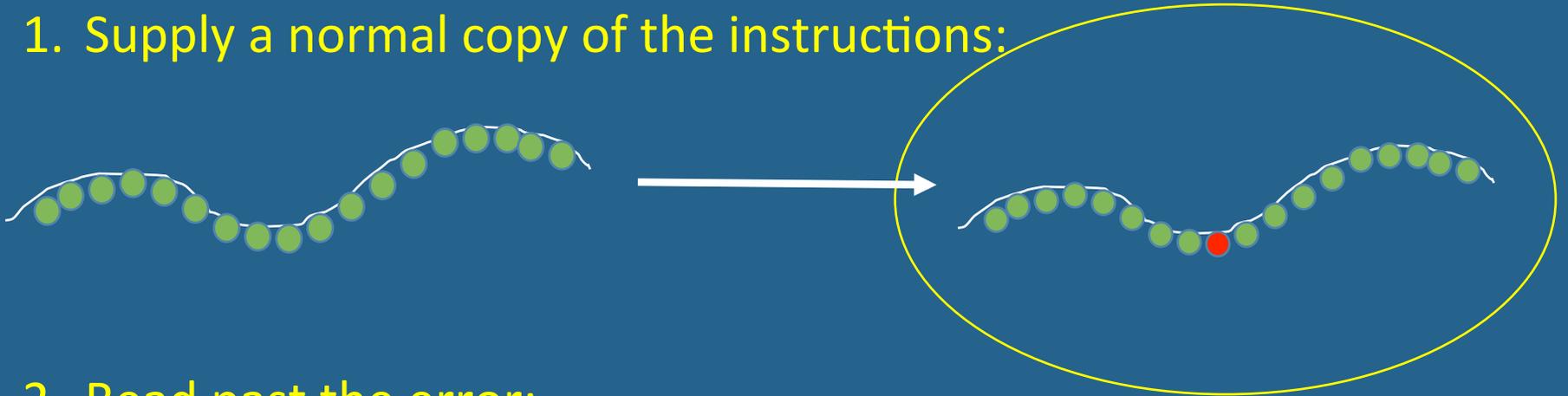
Goal of gene therapy:



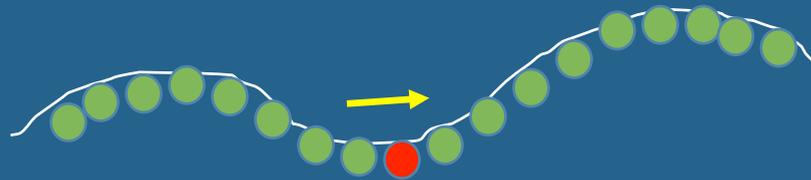
# How does gene therapy work?

Therapies targeting a genetic error could take several forms

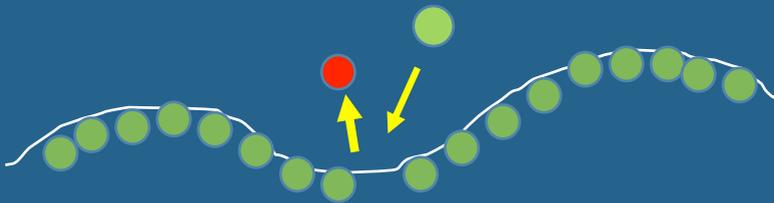
1. Supply a normal copy of the instructions:



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?

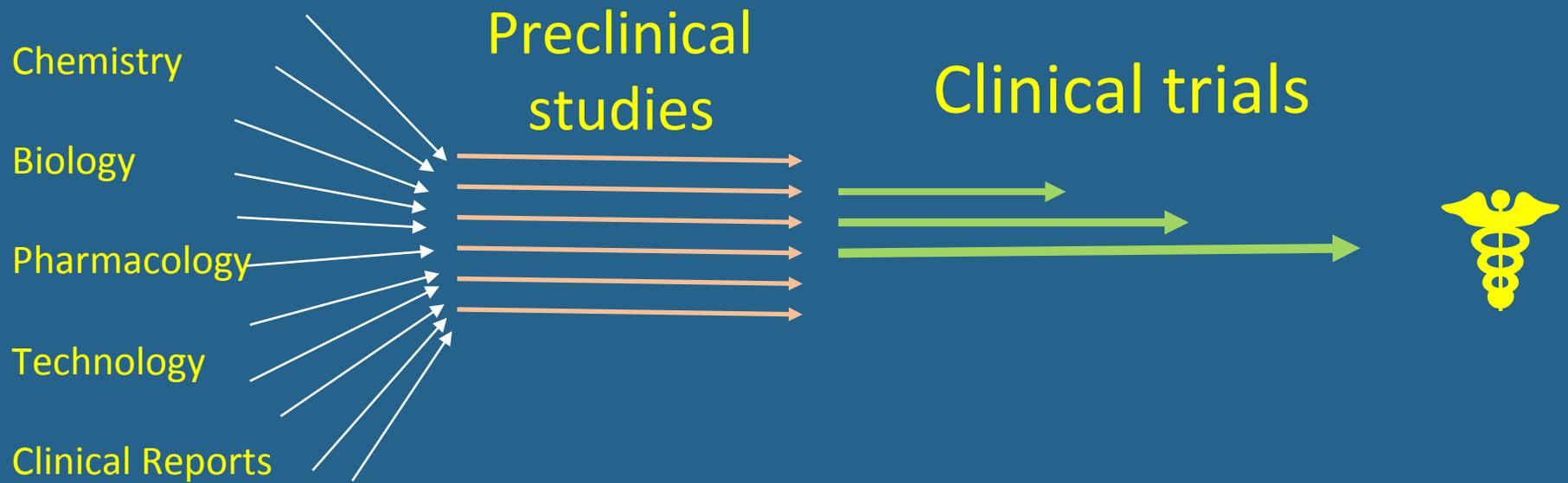
- \*People who have a genetic disorder for which the causative gene has been identified

**VERY IMPORTANT TO HAVE A GENETIC DIAGNOSIS!**

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

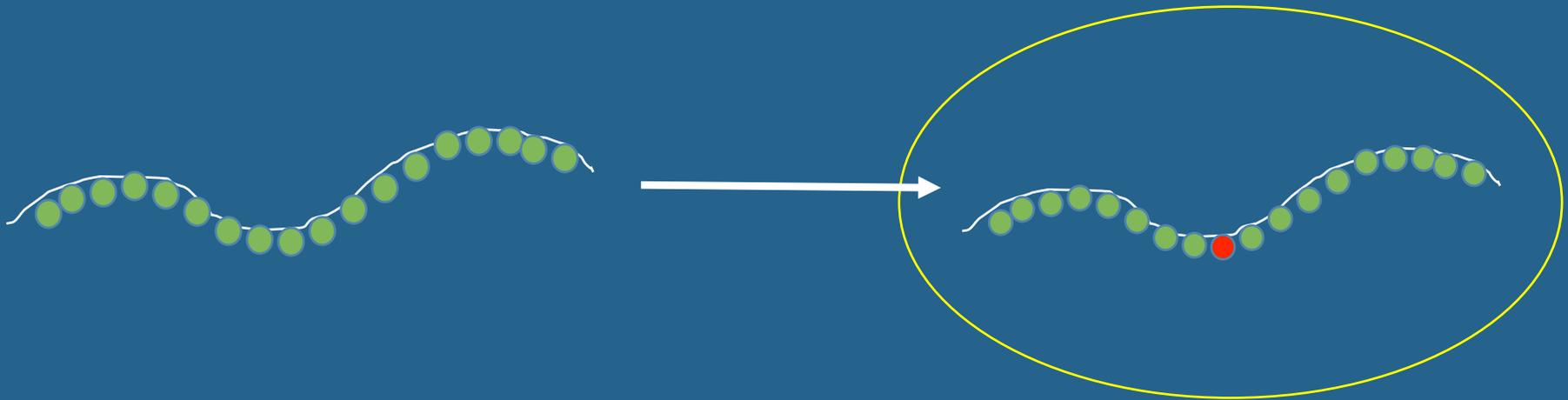
**STAGE OF DISEASE MUST BE KNOWN AND MONITORED**

# 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



This is a gene replacement therapy designed to treat vision loss in USH1B patients

Treatment availability in the future will depend on:

- progress in preclinical research
- success of clinical trials

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](http://www.usher-syndrome.org/take-action/get-genetic-testing.html)

**-Monitor your vision**

[www.usher-registry.org/en/find an expert](http://www.usher-registry.org/en/find_an_expert)

**-Stay connected via the USH Trust and Blue Book**

[www.usher-registry.org](http://www.usher-registry.org)

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USHER SYNDROME  
COALITION