## DNA Mutation Detector

Presented by :- Bacteriophagians

# Topics of Discussion

- DNA Genetic Disorder
- Gene Therapy



## Problems to solve

Detecting the presence of DNA sequence responsible for DNA genetic disorder and stating the type of disorder

Gene Therapy to use to treat the person having DNA disorder.

### Literature review

## DNA DISORDER

- Researchers have made dramatic inroads into the study of polygenic and other complex human diseases, due in large part to knowledge of the human genome sequence, the generation of widespread markers of genetic variation, and the development of new technologies that allow investigators to associate disease phenotypes with genetic loci.
- A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Genetic disorders can be caused by a mutation in one gene (monogenic disorder), by mutations in multiple genes (multifactorial inheritance disorder), by a combination of gene mutations and environmental factors, or by damage to chromosomes (changes in the number or structure of entire chromosomes, the structures that carry genes).
- Although polygenic diseases are more common than single-gene disorders, studies of monogenic diseases provide an invaluable opportunity to learn about underlying molecular mechanisms, thereby contributing a great deal to our understanding of all forms of genetic disease.

## Literature review

#### **GENETHERAPY**

- Background: In 1989, the concept of human gene therapies has emerged with the first approved human gene therapy trial of Rosenberg et al. Gene therapies are considered as promising therapies applicable to a broad range of diseases.
- Methods: A publicly available database, 'Gene Therapy Clinical Trials Worldwide', was used to extract descriptive data on gene therapy clinical trials: (1) number of trials per year between 1989 and 2015; (2) countries; (3) diseases targeted by gene therapies; (4) vectors used for gene delivery; (5) trials status; (6) phases of development.
- Results: Between 1989 and 2015, 2,335 gene therapy clinical trials have been completed, were ongoing or approved (but not started) worldwide. The number of clinical trials did not increase steadily over time; it reached its highest peak in 2015 (163 trials). Almost 95% of the trials were in early phases of development and 72% were ongoing. The United States undertook 67% of gene therapy clinical trials. The majority of gene therapies clinical trials identified targeted cancer diseases.

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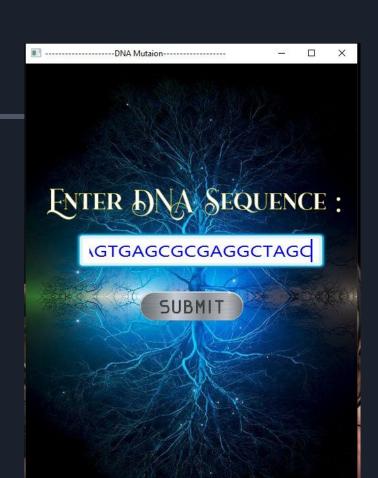
## **DNA** genetic disorder

- It is the disorder caused by expanding nucleotide repeats.
- Where it includes:
  - Finding the presence of repeated sequences through analyzing
     the DNA sequence that causes genetic disorder.
  - And counting the number of repeated sequences occurring in the DNA sequence.
- It happens when the existence of repeated sequence exceeds normal range and enters the disorder Range which results in a particular disease.

## Gene Therapy

- After determining the genetic disorder in a person, gene therapy is used to treat it.
- To normalise the genetic disorder, new DNA is introduced in patient to treat the disorder.
- New DNA contains a functional gene to correct the effects of disease.
- In this we'll insert new DNA sequence into old DNA sequence and then check if it will correct the effect of disorder.
- There are two types of Gene Therapy:-
  - Somatic Gene Therapy- Effects of gene therapy will not passed onto its child
  - <u>Germline Gene Therapy</u>-Effects of gene therapy will passed onto its children.
- This will only be successful if the new DNA does not damage the patient's body.

## RESULTS







## **CONTRIBUTION**

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KUNAL (2019430):- GUI and some parts of code

ABHISHEK JANGID (2019402):- Algorithm and coding using Java.

ADITYA KUMAR JHA (2019404):- Collected data of DNA sequences with genetic disorder from internet and did the exploratory work.

Bhaskar and Dev: maintenance of ppt Aditya Rana: research work

## Thank you.

#### References:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5328344/

https://www.nature.com/scitable/topicpage/rare-genetic-disorders-learning-about-genetic-disease-979/

