

1. Introduction

- Computational biology and gene therapy are making revolutionized advancements for the treatment of many inborn disorders.
- Hemophilia A is among those genetic blood disorders wherein a person bleeds abnormally due to the absence of blood coagulating protein called as Factor VIII.
- Current treatment for hemophilia A patients involves regular self-infusion of concentrates of exogenously derived FVIII which is costly and requires to be infused frequently after 2-3 days.
- Genome editing can be used as a tool to precisely target and repair a genetic defect.
- CRISPR Cas9 is one of the genome editing tool which cuts DNA at mutation and DNA is repaired.

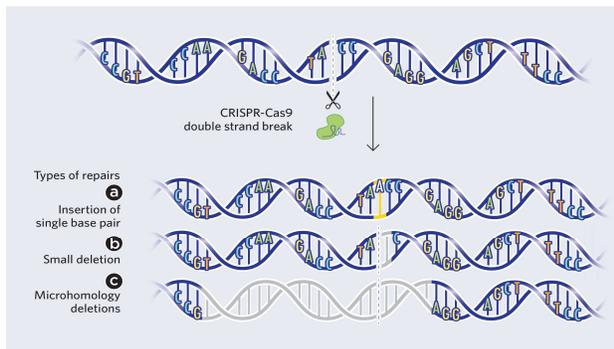


Figure 1 : Types of DNA repair

Source: <https://www.the-scientist.com/the-literature/could-ai-make-gene-editing-more-accurate-65781>

2. Research Objective

- To examine whether machine learning model can predict the type of repair in DNA mutation for hemophilia A patients.

3. Literature Review

- Previous research on genetic mutation identification and classification is carried out on neurological diseases.
- Traditional machine learning and deep learning algorithms are applied to study various mutations in DNA.
- Machine learning model is devised to repair mutations in DNA but it lacks high prediction accuracy.
- In our research, we can investigate on the implementation of various deep learning algorithms and framework to achieve high accuracy in prediction of DNA repair for hemophilia population.

4. Research Methodology

- This research focuses on data containing DNA sequences of hemophilia A population obtained from CRISPR tool.
- Identifying patterns occurring in different types of DNA mutations.
- Making model learn the type of repair essential for specific mutation type.
- Devising a model with higher accuracy is required which can outperform prior research implementations.

5. Technologies

- Machine learning: Python, Scikit learn.
- Visualization: Altair, ggplot
- Deep Learning: Tensorflow, Keras
- Seq: A language build on top of python specially designed for computational biological applications such as DNA sequencing, finding mismatch in DNA base pairs and DNA alignment.

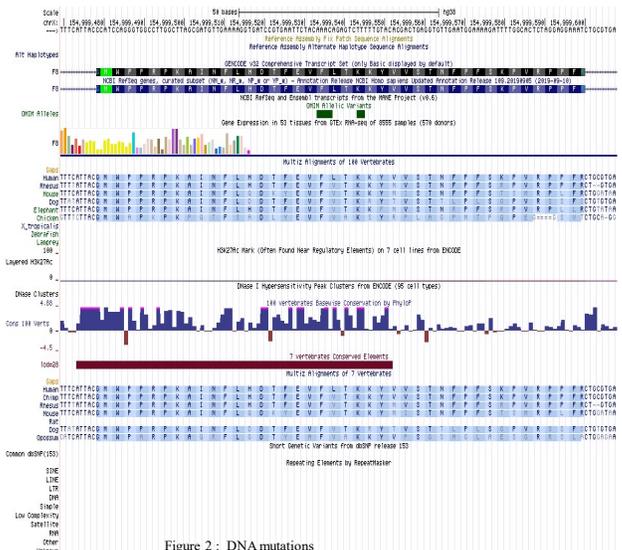


Figure 2 : DNA mutations

Source: <https://genome-euro.ucsc.edu/>

6. Next Steps

- On achieving high precision model, scientist/researcher can utilize it for treatment of hemophilia A with gene editing by targeting exact mutation location.

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