ALPHAGENOME – SCIENECE & TECHNOLOGY

NEWS: Google DeepMind has launched **AlphaGenome**, a powerful AI model designed to predict how genetic mutations impact our health and may even potentially provide new treatments.

- DeepMind CEO **Demis Hassabis** recently won the **Nobel Prize in Chemistry** for work related to genetics and DNA.

WHAT'S IN THE NEWS?

What is the Human Genome?

- The **human genome** refers to the **entire set of DNA** present in a human cell, which contains all the **genetic instructions** necessary for the development, functioning, growth, and reproduction of the organism.
- It acts as a **biological instruction manual**, guiding how cells behave, what proteins they produce, and how the body responds to internal and external stimuli.
- Although most of the genome is identical among all humans, **small variations in DNA sequences (called genetic variants)** can significantly affect an individual's traits, disease susceptibility, and interaction with the environment.
- **Decoding and interpreting the functions of these variations**—especially those in non-coding regions—is one of the most complex and ongoing challenges in genetics and molecular biology.

What is AlphaGenome?

- **AlphaGenome** is a **new artificial intelligence model developed by Google DeepMind** aimed at predicting how genetic mutations influence DNA regulatory functions.
- Unlike earlier models focused mainly on **protein-coding regions**, AlphaGenome provides insights into **non-coding DNA regions**, which do not produce proteins but play crucial roles in **gene expression regulation**.
- It builds upon DeepMind's previous models such as **Enformer**, which predicts gene expression, and **AlphaMissense**, which classifies the potential harm of protein-coding mutations.
- The model is designed to **analyze and predict how mutations affect DNA's control systems**, helping scientists better understand gene regulation on a genome-wide scale.

How AlphaGenome Works

- AlphaGenome processes **long DNA sequences, up to 1 million base pairs in length**, which allows it to evaluate broad genomic contexts.

- The model makes predictions about **thousands of molecular features** that reflect DNA regulatory activity across different tissues and conditions.
- It compares the molecular properties of **normal (unmutated) DNA sequences** with those that carry **specific mutations**, scoring the impact of each variant on gene regulation.
- Some key regulatory features predicted by AlphaGenome include:
    - **Transcription start and stop sites** in various tissues and cell types.
    - **Splicing sites**, where genes are edited to produce different protein forms.
    - **Levels of RNA production**, indicating gene activity.
    - **Accessibility of DNA bases** (i.e., how open the chromatin is for protein binding).
    - **Proximity and binding of specific regulatory proteins** to DNA.

Applications of AlphaGenome

- **Disease Understanding**: Helps identify **potential disease-causing mutations** by predicting their impact on gene regulation and function.
- **Synthetic Biology**: Can be used to **design synthetic DNA sequences** with custom regulatory properties, useful in gene therapies and engineered biological systems.
- **Fundamental Research**: Supports **basic genomic research** by helping scientists map and interpret the **regulatory architecture** of genes in different cell types and tissues.

GENE VERSUS GENOME

GENE	GENOME
A gene is a part of a DNA molecule	The genome is the total DNA in a cell
Hereditary element of genetic information	All set of nuclear DNA
Encodes protein synthesis	Encodes both proteins and regulatory elements for protein synthesis
Length is about a few hundreds of bases	Length of the genome of a higher organism is about billion base pairs
A higher organism has about thousands of genes	Each organism has only one genome
Variations of the gene named alleles can be naturally selected	Horizontal gene transfer & duplication cause large variations in the genome

Limitations of AlphaGenome

- **Long-Range Regulatory Interactions**: The model has limited ability to accurately capture the influence of **regulatory elements located over 100,000 base pairs away** from the target gene.
- **Cell- and Tissue-Specific Accuracy**: The model needs improvement in precisely predicting **regulatory effects that are unique to specific cell types or tissues**.
- **Not Personalized**: AlphaGenome is **not designed or validated to interpret individual genomes**, meaning it's not ready for direct clinical or personal genetic analysis.
- **Incomplete Trait and Disease Predictions**: While it can predict **molecular consequences** of mutations, it doesn't integrate **complex environmental or biological factors**, which are essential to fully understand human traits or disease risks.

Source: https://www.indiatoday.in/technology/news/story/google-deepmind-unveils-alphagenome-ai-to-decode-how-dna-changes-impact-human-health-2746597-2025-06-26