BP-02: Artificial intelligence annotation of gene variants linked to rare

BAADECHE, Abdelhadi[1], CHEHILI, Hamza[2], ZIDOUNE, Housna[3].

1, 2, 3 Microbiological Engineering and Applications Laboratory, Constantine 1- Frères Mentouri UniversityAlgeria

Email: ***abdelhadi.baadeche@doc.umc.edu.dz***

Subject description: Rare diseases, also referred to as orphan diseases, are medical conditions characterized by their low prevalence within a specific population (generally 1 in 10.000).

The rarity of these diseases presents challenges in terms of accurate diagnosis, availability of effective treatments, and adequate research funding. These conditions encompass a diverse range of complex disorders caused by genetic mutations.

Artificial intelligence (AI) plays a vital role in rare disease annotation using its data analysis, pattern recognition and knowledge integration capabilities

Objectives: Develop or use an AI-powered tool to accurately annotate and prioritize genetic variants associated with a particular rare disease, aiding researchers, clinicians, and geneticists in understanding the disease's genetic basis and potential treatment options.

Methods: Collect and integrate relevant genetic databases, medical literature, andclinical trial data related to the specific rare disease.

Implement machine learning algorithms to identify and classify genetic variants frompatient data and reference genomes

Results and discussion: The use of these tools accelerates the identification andprioritization of relevant genetic variants, reducing the time needed for manual analysis.

Conclusion: Having observed the positive outcomes resulting from the use of AI approach, it prompt us to develop our work further and try to create an advanced AI solutionthat significantly contributes to annotating genetic variants responsible for specific rare diseases.

Keywords: Artificial intelligence, Rare diseases, Genetic database.