



## BP-07: The application of machine learning techniques in rare diseases.

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**Subject description:** Emerging machine learning (ML) techniques have the potential to greatly improve rare disease (RDs) research and treatment. The use of artificial intelligence (AI) technologies can be especially advantageous for the study of RDs, which are a diverse group of diseases that impact a small percentage of the whole population and significantly underrepresented in basic and clinical research. The difficulties faced by RDs (such as small patient population, geographical dispersion, low diagnostic rates, etc.) can be overcome by using ML techniques.

**Objectives:** This review aims to highlight the accomplishments of AI algorithms in the study of rare diseases and to guide researchers on which strategies have proven to be the most beneficial.

**Methods:** The study will focus on a few rare diseases. The Orphanet categorization was used, and only RDs with Orpha codes were considered. And will look at which AI methodologies have been most successful in their research.

**Results and discussion:** ML techniques demonstrate that no single strategy excels universally; success is dependent on unique tasks and resources. The complexity, interpretability, and data requirements of models differ. While deep learning can capture complicated patterns, it may be difficult to interpret, as opposed to simpler models such as logistic regression. There is a clear trade-off between model complexity and performance. Ensemble learning, like random forests, is resistant to noisy data. Deep learning necessitates enormous computational resources. Tuning hyperparameters is crucial, and technique selection should be guided by domain-specific factors.

**Conclusion:** In conclusion, from the standpoint of precision medicine, AI algorithms can help to design individualized treatment plans by finding biomarkers linked with a specific rare disease. AI systems that discover, forecast, and classify mutations can advance RDs diagnosis, raising these figures and uncovering new disease causes and therapeutic targets.

The AI-mediated knowledge of RDs could considerably accelerate therapeutic development.

**Keywords:** artificial intelligence; machine learning; rare diseases; diagnosis; precision medicine.