



## BP-09: Decoding the Genome: Advancing Anomaly Detection through Machine Learning

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**Subject description:** This presentation explores the fusion of genomic analysis and machine learning with the aim of revolutionizing anomaly detection in genetics. This advancement is seen as propelling precision medicine and enhancing advanced diagnostics.

**Objectives:** The objectives include the investigation of the application of machine learning in the detection of genetic anomalies. This aims to elucidate its potential in early disease identification and the provision of personalized healthcare.

**Methods:** The presentation begins with an introduction to genomics, highlighting the necessity of artificial intelligence in dealing with the vast amount of genomic big data. It then proceeds to delve into various machine learning tools, such as DeepVariant, VarSome Clinical, and Deep SEA. Throughout this exploration, the presentation unveils the data sources, predictive capabilities, and the profound impact these tools have on the interpretation of genomics.

**Results and discussion:** During this segment, it is demonstrated that by harnessing the prowess of artificial intelligence, enhanced accuracy in the identification of genetic anomalies can be showcased. This results in the faster analysis of vast genomic datasets, opening the door to potential groundbreaking biomedical discoveries.

**Conclusion:** In conclusion, the amalgamation of genomics and machine learning heralds a paradigm shift in the domains of disease detection and treatment, ushering in a new era characterized by tailored healthcare.

**Keywords:** Genomic Anomalies, Machine Learning, Precision Medicine, Early Disease Detection, Genomic Interpretation