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Abstract Topic:- Complex traits and polygenic disorders

Abstract Title:- A deep learning-based risk assessment tool for predicting Parkinson's disease genetic risk

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**Aims:**-To Leverage power of deep neural networks using high dimensional genomic data and develop a Deep learning model which predicts genetic risk of Parkinson's Disease (PD).

**Methods:-** We developed deep neural network models for predicting genetic risk for PD and evaluated the performance against conventional weighted Polygenic risk scoring framework. For this we have used a cohort of 660 PD patients and 1363 controls. We stratified individuals by applying a threshold on the probability score from a trained neural network model.

**Results:-** We observed that Deep Neural Network based models outperformed conventional PRS method in terms of odds-ratios in the highest decile bin (OR.Deep.PRS =7.28) vs (OR.PRS=1.68) and ROC-AUC in our validation study.Our approach identified 51% individuals as high risk as compared to 32% using conventional method.

**Conclusions:-** The Parkinson's disease genetic susceptibility has been previously described by ~1400 common variants. It has been shown that the Polygenic Risk Score (PRS) has been successful in predicting genetic risk of developing PD. However, because of the complexity of genomic data, the present statistical framework cannot comprehensively capture the genetic risk of PD. Methods such as deep learning capture nonlinearity within high-dimensional genomic data, may enable more efficient disease risk prediction compared to the conventional PRS approaches. Our results suggest that deep learning methods are highly efficient and are better tools for assessment of genetic risk in PD and other diseases, identifying high genetic risk individuals which can be used as a risk screening tool.

**Keywords:-** Parkinson's disease, Deep learning, Polygenic Risk scores, Genetic-risk prediction tool, Neural Networks