Abstract

Hereditary ataxias are a group of heterogeneous diseases with more than 50 subtypes arising from different genes. Each disease can only be diagnosed with a specific test or group of tests. The diseases differ slightly from each other in clinical features as well as in progression. Only an expert geneticist can distinguish subtypes phenotypically provided patient has complete clinical picture. This diagnostic problem is addressed using a clinical decision support system that narrow down the list of probable diagnoses when given the phenotypic data of a patient. The system uses a database of clinical features of disease subtypes to calculate the posterior probability of having the diseases when given with a phenotype profile. Due to scarcity of patients from each subtype, to develop a database with real values, a database was created using probability of symptoms and signs given in medical literatures as qualitative expressions transformed in to quantitative values and used in the database. Probability calculations were done applying the Bayesian theory for each symptom separately. Average of the resulted values for each disease was taken as a probability score and diseases were listed from highest probability score to the lowest. The appropriate genetic tests were then suggested by the system as outcome.