# Inheritance Pattern Prediction – An Ophthalmic Model for Digital Pedigree Feature Extraction and Machine Learning

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**Purpose**: For conditions with genetic heterogeneity, determining the pattern of inheritance may require complex pedigree interpretation. However, non-genetics providers may not feel confident performing this task, and not all patients have access to genetic counseling services. Therefore, a computer program has been developed to directly extract information about a patient's family history from a digital version of their pedigree, and a machine-learning algorithm has been created whose input is this extracted data and whose output is the most likely pattern of inheritance.

**Methods**: The pedigrees of patients from the Kellogg Eye Center Retinal Dystrophy Clinic for whom the genetic cause of disease had been identified were analyzed with 3 methods: 1) inheritance pattern prediction by genetic counselor-trained students (277 patients), 2) clinician-reported answers to 12 questions about pedigree structure (100 patients), and 3) computer-calculated answers to the same questions using feature extraction, with tolerance for user input errors, on a digitalized patient pedigree (90 patients). The 3 cohorts have an overlap of 70 patients. A machine learning model was used on the second two methods to predict the pattern of inheritance. Results were compared with the pattern of inheritance associated with the genetic diagnosis for that patient.

**Results**: Student-predicted pattern of inheritance had 84% accuracy, machine learning on human answers to questions had 80% accuracy (SD 7.5%), and machine learning on computer-calculated answers to the questions had 76% accuracy (SD 9.8%). Our machine-learning algorithms provided results with similar accuracy to that of humans, and this algorithm will likely improve over time and with the addition of more data.

**Conclusions**: Machine learning is becoming more common in medicine, and it is a useful tool for examining the pattern of inheritance for genetic conditions. This may be helpful for determining appropriate genetic testing panels to order and counseling about risk calculation for relatives when genetics providers are unavailable.