Development of a Protocol for Simultaneously Sequencing Multiple Inherited Retinal Diseases' Founder Mutations

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Inherited retinal diseases may cause retinal degeneration which can lead to blindness. There are about 250 different genes associated with inherited retinal diseases resulting in many different possible mutations and genes involved in the same disease. The large number of mutations makes the genetic diagnosis complicated, thus long and inefficient. In the present study we purpose a new protocol in which a simultaneous sequencing of many mutations makes this process more efficient. In order to develop this protocol 50 inherited retinal diseases' founder mutations were chosen and special primers for combined multiple PCR were designed. DNA from patients with retinal diseases was purified and sequenced by next generation sequencing. This process was carried out several times using different DNA samples while calibrating the protocol each time. Using this protocol, the genetic cause for 42 patients out of 225 patients (18%) was found for the first time. These results show that the protocol is successful and the genetic cause for new patient could be found by using it. We achieved the goal of developing an efficient protocol in terms of time and cost. This protocol can be implemented regularly in the lab as a way to improve the performance of genetic diagnosis initial process. In the future, this protocol can also be used for other genetic diseases and different research purposes such as sequencing many new regions in the DNA in order to search new mutations.

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