Title: A novel mutation in exon 39 of *PRPF8* is responsible for RP with variable expressivity

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Purpose: To find the gene mutated in a large English family affected by autosomal dominant Retinitis Pigmentosa (adRP) with variable expressivity.
Methods: After examination, blood samples were obtained for ten members of this family. Genomic DNA was isolated and markers for know adRP genes were tested. RNA was extracted and cDNA made for an affected individual. cDNA for *PRPF8* was sequenced for this individual as well as direct sequencing was performed on the DNA for the reminding individuals.
Results: The disease in the family was linked to chromosome 17, at the *PRPF8* locus. A new change 6445 C>T was found in exon 39 of *PRPF8*. The change segregates with the disease in the family. It was not found in 130 controls indicating this change is responsible for the disease in the family. It is interesting to note that this is the first time a mutation in *PRPF8* is found in another exon than exon 43.
Conclusions: A new mutation Ser 2118 Phe was reported in exon 39 of *PRPF8* in a family affected by adRP with variable expressivity. Incomplete penetrance can be considered as an extreme case of variable expressivity. It can be difficult to distinguish between them at young age. So far, only mutations in *PRPF31* have been discovered in adRP families with incomplete penetrance. We suggest that it will be worth screening *PRPF8* in adRP families with incomplete penetrance and when *PRPF31* mutation is absent.