Supplementary Figure S1

We performed an extensive review of the literature (PubMed: accessed April 12, 2018) to identify all known mutations in the RP1 gene (including those reported herein) that have been associated with hereditary retinal dystrophies.

Schematic representation of the mutations in the RP1 gene and their location. Mutations that have been associated with autosomal dominant RP are indicated left from the gene, whereas mutations that have been associated with autosomal recessive RP, MD or CRD are indicated on the right side. The portion of the gene that encodes the doublecortin (DCX) and a region homologous to the Drosophila melanogaster bifocal (BIF) domain, are indicated with the striped and checkered pattern, respectively. Mutations indicated with an arrowhead represent novel mutations that are identified in this study.

† Missense mutation with uncertain pathogenicity.

The mutations can be divided into 6 groups: group 1 mutations (in grey) reside in a hotspot region in exon 4 and are expected to result in a truncated protein with dominant-negative activity; group 2 (orange) includes protein-truncating mutations located in exon 2 and 3, whose transcripts are thought to be subject to nonsense mediated decay (NMD); group 3 (red) encompasses protein-truncating mutations in the proximal part of exon 4; group 4 (green) includes mutations in exon 4 which, considering their location, could lead to autosomal dominant RP, yet they are not pathogenic in a heterozygous state; group 5 (purple) includes protein-truncation mutations in the distal part of exon 4 with residual function; group 6 (black) contain missense variants.