Supplementary Table S1. Subpanel of 74 genes associated with retinitis pigmentosa, choroideremia, macular dystrophy, cone-rod dystrophy, Leber congenital amaurosis, congenital stationary night blindness, and Usher syndrome. In bold: 24 out of 28 genes associated with adRP (RetNet) that were screened by NGS.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Gene</th>
<th>Gene</th>
<th>Gene</th>
<th>Gene</th>
<th>Gene</th>
</tr>
</thead>
<tbody>
<tr>
<td>ABCA4</td>
<td>CNGB1</td>
<td>KLHL7</td>
<td>PRPF6</td>
<td>RPGIP1</td>
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<td>CRB1</td>
<td>LCA5</td>
<td>PRPF8</td>
<td>SAG</td>
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<tr>
<td>ADAMTS18</td>
<td>CRX</td>
<td>LRAT</td>
<td>PRPH2</td>
<td>SEMA4A</td>
<td></td>
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<td>AIPL1</td>
<td>CYP4V2</td>
<td>MAK</td>
<td>RBP3</td>
<td>SNRN200</td>
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<td>DHDDS</td>
<td>MERTK</td>
<td>PD1</td>
<td>SPATA7</td>
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<tr>
<td>BEST1</td>
<td>EYS</td>
<td>NMNAT1</td>
<td>RDH5</td>
<td>TOPORS</td>
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<tr>
<td>C1QTNF5</td>
<td>FAM161A</td>
<td>NR2E3</td>
<td>RDH12</td>
<td>TTC8</td>
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<tr>
<td>C2orf71</td>
<td>FSCN2</td>
<td>NRL</td>
<td>RGR</td>
<td>TULP1</td>
<td></td>
</tr>
<tr>
<td>C8orf37</td>
<td>GUCA1B</td>
<td>OFD1</td>
<td>RHO</td>
<td>USH2A</td>
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</tr>
<tr>
<td>CA4</td>
<td>GUCY2D</td>
<td>PDE6A</td>
<td>RBP1</td>
<td>ZNF513</td>
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<tr>
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<td>HK1</td>
<td>PDE6B</td>
<td>ROM1</td>
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<tr>
<td>CEP290</td>
<td>IDH3B</td>
<td>PDE6G</td>
<td>RP1</td>
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<tr>
<td>CERKL</td>
<td>IMPDH1</td>
<td>PRCD</td>
<td>RP2</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CHM</td>
<td>IMPG2</td>
<td>PROM1</td>
<td>RP9</td>
<td></td>
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<tr>
<td>CLRN1</td>
<td>IQCB1</td>
<td>PRPF3</td>
<td>RPE65</td>
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<tr>
<td>CNGA1</td>
<td>KCNJ13</td>
<td>PRPF31</td>
<td>RPGR</td>
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Supplementary Table S2. Most frequent pathogenic variants found in our population.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Exon</th>
<th>Nucleotide change</th>
<th>Protein change</th>
<th>No. Families (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>RHO</td>
<td>5</td>
<td>c.1040C&gt;T</td>
<td>p.(Pro347Leu)</td>
<td>10 (3.9)</td>
</tr>
<tr>
<td>NR2E3</td>
<td>2</td>
<td>c.166G&gt;A</td>
<td>p.(Gly56Arg)</td>
<td>7 (2.7)</td>
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<tr>
<td>RHO</td>
<td>2</td>
<td>c.403C&gt;T</td>
<td>p.(Arg135Trp)</td>
<td>5 (1.9)</td>
</tr>
<tr>
<td>C1QTNF5</td>
<td>15</td>
<td>c.489C&gt;A</td>
<td>p.(Ser163Arg)</td>
<td>4 (1.6)</td>
</tr>
<tr>
<td>PRPF3</td>
<td>11</td>
<td>c.1481C&gt;T</td>
<td>p.(Thr494Met)</td>
<td>3 (1.2)</td>
</tr>
<tr>
<td>RHO</td>
<td>2</td>
<td>c.491C&gt;G</td>
<td>p.(Ala164Glu)</td>
<td>3 (1.2)</td>
</tr>
<tr>
<td>RHO</td>
<td>3</td>
<td>c.544G&gt;A</td>
<td>p.(Gly182Ser)</td>
<td>3 (1.2)</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td></td>
<td></td>
<td>35 (13.6)</td>
</tr>
</tbody>
</table>
Supplementary Figure S1. Pedigrees of the reclassified families. Cosegregation in available family members are shown. m: mutated allele. wt: wild-type allele.

**LORD: late-onset retinal degeneration**
m: C1QTNF5 Ex.15 c.489C>A p.(Ser163Arg)

**RP-2760**

**RP-2409**

**RP-0488**

**RP-0911**
RP-0858
m: RPGR Ex.6 c.485_486del p.(Phe162Tyrfs*4)

RP-0615
m: RPGR Ex.15 c.2405_2406del p.(Glu802Glyfs*32)

RP-2646
m: RPGR Ex.15 c.2296_2299del p.(Gly766Asnfs*48)

RP-2683
m: RPGR Ex.8 c.888_889del p.(Ile297Lysfs*48)

X-linked genes
**RP-1313**

m: RP2 Ex.1 c.1A>G p.(Met1?)

**RP-1682**

m: RP2 Ex.1 c.14_16del p.(Phe5del)

**RP-2204**

m: CHM Ex.5 c.340G>T p.(Glu114*)

**RP-2518**

m: CHM Ex.6 c.757C>T p.(Arg253*)
RP-1123
m1: RDH5 Ex.4 c.625C>T p.(Arg209*)
m2: RDH5 Ex.5 c.776C>T p.(Pro259Leu)

RP-0038
m1: CNGA1 Ex.5 c.94C>T p.(Arg32*)
m2: CNGA1 Ex.6 c.131delA p.(Glu44Glyfs*49)

RP-1217
m: EYS IVS28 c.5928-2A>G

RP-1446
m1: USH2A Ex.13 c.2299del p.(Glu767Serfs*21)
m2: USH2A Ex.62 c.12094G>A p.(Gly4032Arg)

RP-1455
m1: ABCA4 Ex.19 c.2888del p.(Gly963Alafs*14)
m2: ABCA4 Ex.48 c.6688del p.(Leu2230Serfs*17)
m3: ABCA4 Ex.8 c.950del p.(Gly317Alafs*57)

Autosomal recessive RP genes
Supplementary Figure S2. Haplotype analysis using 3 microsatellite markers with high heterozygosity (D19S606, D19S596, and D19S879) flanking 1.5 Mb around the CRX gene and an intragenic polymorphic marker (D19S902) in RP-1092 and RP-1192 families with the deletion of exons 3 to 4 in the gene. Haplotype analysis suggests a recurrent mutation. m: mutated allele (deletion of exons 3 to 4 of the CRX gene). wt: wild-type allele.

<table>
<thead>
<tr>
<th>Markers</th>
<th>Start (Mb)</th>
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<tbody>
<tr>
<td>D19S606</td>
<td>47.97</td>
</tr>
<tr>
<td>D19S902</td>
<td>48.33</td>
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<tr>
<td>Del E3-4</td>
<td>48.34</td>
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<tr>
<td>D19S596</td>
<td>49.25</td>
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<tr>
<td>D19S879</td>
<td>49.52</td>
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CRX: chr19:48325097-48346586
Supplementary Table S3. Percentage of characterization and prevalence of the *RHO*, *PRPF31*, *PRPH2*, and *RP1* mutations in different adRP populations. Char.: characterization. No.: number. * Various techniques, including NGS.

<table>
<thead>
<tr>
<th>Population</th>
<th>% Char.</th>
<th>No. adRP cases</th>
<th>No. genes screened</th>
<th>Technique</th>
<th>%RHO</th>
<th>%PRPF31</th>
<th>%RP1</th>
<th>%PRPH2</th>
<th>Reference</th>
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<tbody>
<tr>
<td>Spanish</td>
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<td>258</td>
<td>-</td>
<td>Various *</td>
<td>20.9</td>
<td>8.1</td>
<td>4.3</td>
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<td>Present study</td>
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<tr>
<td>American</td>
<td>78.5</td>
<td>270</td>
<td>-</td>
<td>Various *</td>
<td>30.7</td>
<td>8.9</td>
<td>4.8</td>
<td>7</td>
<td>Daiger et al., 2014(^1)</td>
</tr>
<tr>
<td>Belgian</td>
<td>56</td>
<td>86</td>
<td>-</td>
<td>Various *</td>
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<td>10.5</td>
<td>10.5</td>
<td>4.7</td>
<td>Van Cauwenbergh et al., 2017(^2)</td>
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<tr>
<td>French Canadian</td>
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<td>9</td>
<td>Sanger</td>
<td>18.3</td>
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<td>5</td>
<td>Coussa et al., 2015(^3)</td>
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<tr>
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<td>3.9</td>
<td>5.2</td>
<td>Oishi et al., 2014(^4)</td>
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<tr>
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<td>Sanger</td>
<td>16</td>
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<td>5</td>
<td>-</td>
<td>Ziviello et al., 2005(^5)</td>
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<tr>
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<td>-</td>
<td>Yang et al., 2014(^6)</td>
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<tr>
<td>South African</td>
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<td>SSCP</td>
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<td>-</td>
<td>-</td>
<td>Roberts et al., 2000(^7)</td>
</tr>
<tr>
<td>Indian</td>
<td>2</td>
<td>53</td>
<td>4</td>
<td>Sanger</td>
<td>-</td>
<td>2</td>
<td>-</td>
<td>-</td>
<td>Gandra et al., 2008(^8)</td>
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</tbody>
</table>
References: