

RPGR ORF15

RPGR ORF15 is arguably one of the most difficult to sequence coding regions in the human genome. Unfortunately, most of the mutations in *RPGR* are in the region (usually frameshift mutations). This region cannot be covered by most generic methods. Since mutations in *RPGR* can be found in isolated cases (with no family history indicating X-linked inheritance), sequencing *RPGR* ORF15 becomes essential in order to have a comprehensive and thorough analysis.

At Molecular Vision Laboratory, we have developed a unique method to sequence ORF15. The region is first enriched by long range PCR and then sequenced by Illumina Nextera XT and MiSeq. We have validated the in-house developed method by sequencing ~ 200 samples tested elsewhere. The result will soon be submitted to peer-reviewed journal. Our efforts on the sequencing of *RPGR* ORF15 and several other hard-to-sequence regions differentiate our service from many others. In order to reach to 100% mutation detection rate, we will continuously improve our platforms and we will continue to develop necessary and cost-effective methods.

The most difficult to sequence region in *RPGR* ORF15: (hg19), p.800-1030 (Highly repetitive, G & A rich region):

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GAGCAARCAGWGGGARACG
GGAGGAAAAGARGAGCGAGGGKAAGKARAGGGACGGGAAGKRGACGAGRRGWAAGCACA
GACGGAAGAGGAAGAKKAGKAGGGTGAGGS GGAAGAGGAGGARCRGGAGGGGGAAGAGGA
GKARCGRGA RGGGGAAGRGCAGKA AGGAGARGGGAAAGGGAGGAAGAAGGGGARKAAGC
AGAA GCGGAGGRAGAAGGGGAGGARGGAGAAGGGKRGGGGAA GAGGAGGAAGGAKAAGG
GGAGGGAGAAKARGAAGGAGAAGGGGAGKAGAGAAGAGGAGGAAGGAGAAGGGKAGGGRCA
AGAGGAAGGAGAAGRGGAGGGAGAAGAGCAGGAAGGAGAAGGGAAAAGGGAGGAGGAAGG
AGAGGAWKAGAGARGGGAGGGGGAAKRGGAGGAWGGAKAAGGGGAAKGGGAGGADGGAGA
AGGGGAGGGGARKAGCARGAWGGAGAA TGGGAGGGGKAAGAGGRGGAAR GAGAAGGCCA
RGGGGAAGAGGAAGGAGAAAGGKAAAGRRGAGGADKAGAAAGGCAGGGGGAAGAGGAGGA
AGGAGAAGGGGAGGGGGAAGAGGAAGGGAAGGGGAACMAGAAGCCSAGGARGAAGGACAGGG
AGAGCAAGAAGGGCAGGGAGAA GCSGAGKAAGAAGAGGAAGGGCA
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