### Genetics

**Is a DNA sample available?**
- Please select

**Patient genetically solved?**
- Please select

**Inheritance pattern based on the genetic findings**
- Please select

### Causative mutations

**Locus 1 (causative mutation)**
- Gene (HGMD name)
- Mutation 1 (cDNA)
- Mutation 1 (protein)
- Mutation 2 (cDNA)
- Mutation 2 (protein)

**Mutations of uncertain pathogenicity**

**Locus 1 (causative mutation)**
- Gene (HGMD name)
- Mutation 1 (cDNA)
- Mutation 1 (protein)
- Mutation 2 (cDNA)
- Mutation 2 (protein)

### Screened genes

**Screened gene 1**
- Gene (HGMD name)

**Accession no. cDNA**

**Molecular method**
- Genotype microarray (APEX)
- Sanger sequencing
- Targeted NGS: Chromosomal area
- Targeted NGS: Gene set
- Exome NGS: All human genes
- PCR
- Other

**Specify other molecular method**
- APEX microarray; specify array and year; see Asper Ophthalmics.

**Targeted NGS: Chromosomal area**
- Specify hg build and area (e.g. hg19 chr21:33,031,597-33,041,570; see Human (Homo sapiens) Genome Browser Gateway) or flanking RS numbers.

**Targeted NGS: Gene set**

**Used filter: IRD-genes**
- Please select

**Used filter version**
- Please select

**Used enrichment probes**

**Specify used enrichment probes**

**Mapping information**

**Additional information**