

# X-linked juvenile retinoschisis (XLRS)

OMIM 312700

Gene: RS1

Locus: Xp22

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**SERVICE:** mutation and dosage analysis of the RS1 gene

**TESTING:** Diagnostic: clinically affected males  
Presymptomatic: males at risk of developing XLRS (known RS1 mutation)  
Carrier: female relatives of affected males (known RS1 mutation)

**REFERRALS:** from Consultant Clinical Geneticists and Consultant Ophthalmologists only

## TARGET REPORTING TIME AND COSTS

(Non NHS patients are subject to 20% surcharge and payment must be agreed prior to testing)

Diagnostic:	8 weeks	£589	(sequencing + MLPA of exons 1-6)
Presymptomatic:	2 weeks	£195	(sequence one exon)
		£157	(dosage analysis)
Carrier:	2 weeks	£195	(sequence one exon)
		£157	(dosage analysis)

## TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-6 and splice site boundaries of the RS1 gene
- Multiplex ligation dependent probe amplification analysis of exons 1-6 of the RS1 gene

Approximately 75% of mutations are missense, with 86% of all mutations located in exons 4-6 of the gene.

## SAMPLE REQUIREMENTS

- 5ml blood in EDTA or 50ul DNA
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**

Samples should be accompanied by a FULLY completed request card (available from the laboratory). Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital.

## CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

**East Anglian Medical Genetics Service  
Genetics Laboratories  
Molecular Genetics, Box 158  
Level 6, Addenbrooke's Treatment Centre  
Addenbrooke's Hospital  
Cambridge CB2 0QQ**

**Tel: 01223 348866  
Fax: 01223 348870**

**Acting Head of Laboratory: [becky.treacy@addenbrookes.nhs.uk](mailto:becky.treacy@addenbrookes.nhs.uk)**