

Emery Dreifuss muscular dystrophy – X linked (XL-EDMD)

OMIM 310300

Gene: Emerin Locus: Xq28 OMIM: 300384

SERVICE: mutation analysis of the emerin gene

TESTING: Diagnostic: clinically affected males
Carrier: female relatives of clinically affected patients (known emerin mutation)
Prenatal: at risk of having an affected child (known emerin mutation)

*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: from Consultant Clinical Geneticists and Consultant Neurologists

TARGET REPORTING TIME AND COSTS

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

Diagnostic:	8 weeks	£432 (sequencing)
Carrier:	2 weeks	£195 (sequence one exon)
Prenatal:	3 days	£555 (sequence one exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-6 and splice site boundaries of the emerin 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

UKGTN 'Testing criteria' template

Patient name:

Patient address:

Name of referrer:

Title/Position:

Department/Hospital:

Name of Disease/test:

Emery Dreifuss muscular dystrophy, X linked

Referrals only will be accepted from one of the following:

(Please indicate with a tick which category refers to the referrer).

Referrer	Tick if this refers to you.
Consultant Clinical Geneticist	
Consultant Neurologist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:

Criteria	Tick if this patient meets criteria
1) Absence of emerin immunostaining in muscle. If this cannot be performed then all the clinical features below should be present:	
2) Early contractures of Achilles tendons, elbows or spine	
3) Slowly progressive muscle wasting and weakness with predominantly humeral and peroneal distribution	
4) Cardiac conduction defect and/or other evidence of cardiomyopathy	

If the sample does not fulfil these criteria and you still feel that testing should be performed please contact the molecular genetics laboratory