

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: Clinical Geneticists and Neurologists

The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic: 8 weeks

£295 (sequencing)

Carrier: 2 weeks

£145

Prenatal: 3 days

£350 (including maternal contamination studies)

TECHNICAL INFORMATION

• PCR and fluorescent sequence analysis of exons 1-6 and splice site boundaries of the emerin gene
More than 99% of patients with established X-linked inheritance or individuals with no emerin expression as determined by immunodetection studies of muscle tissue have point mutations in the emerin gene.

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- 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
- Samples and paperwork must include three unique and matching patient identifiers

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

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.

CONTACT DETAILS

Genetics Laboratories, Box 143
Level 6, Addenbrooke's Treatment Centre
Addenbrooke's Hospital
Cambridge CB2 0QQ
Tel: +44 (0) 1223 348866
Fax: +44 (0) 1223 348870
Email: becky.treacy@addenbrookes.nhs.uk

Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

| | |
|---------------------|--|
| Name of disease(s): | Emery Dreifuss muscular dystrophy, X-linked |
| Name of gene(s): | Emerin |

| | |
|---------------------------------|----------------|
| Patient name: | Date of birth: |
| Patient postcode: | NHS number: |
| Name of referrer: | |
| Title/Position: | |
| Department/Hospital: | |
| Contact email/telephone number: | |

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 | <input type="checkbox"/> |
| Consultant Neurologist | <input type="checkbox"/> |

Minimum criteria required for testing to be appropriate:

| Criteria | Tick if this patient meets criteria |
|--|-------------------------------------|
| Male with absence of emerin immunostaining in muscle OR if this cannot be performed then all the clinical features below should be present: | <input type="checkbox"/> |
| 1. Early contractures of Achilles tendons, elbows or spine AND | <input type="checkbox"/> |
| 2. Evidence of muscle involvement as elevated serum CK OR slowly progressive muscle wasting and weakness with predominantly humeral and peroneal distribution AND | <input type="checkbox"/> |
| 3. Cardiac conduction defect and/or other evidence of cardiomyopathy OR | <input type="checkbox"/> |
| Obligate female carriers where father satisfies above criteria | <input type="checkbox"/> |
| At risk family members with known mutation | <input type="checkbox"/> |

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