

**THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.**

**PATIENT HISTORY FOR EMERY-DREIFUSS MUSCULAR DYSTROPHY (EDMD) TESTING**

**Patient Name:** \_\_\_\_\_ **Date of Birth:** \_\_\_\_\_ **Sex:**  Female  Male  
**Ordering Provider:** \_\_\_\_\_ **Provider's Phone:** \_\_\_\_\_  
**Practice Specialty:** \_\_\_\_\_ **Provider's Fax:** \_\_\_\_\_  
**Genetic Counselor:** \_\_\_\_\_ **Counselor Phone:** \_\_\_\_\_

**Patient's Ethnicity/Ancestry** (check all that apply)

African American/Black  Asian  Hispanic  White  Other: \_\_\_\_\_

**List country of origin (if known):** \_\_\_\_\_

**Does the patient have clinical findings of EDMD?**  No  Yes (check all that apply and describe)  Unknown

- Joint contractures (age of onset: \_\_\_\_\_)
  - Elbow flexors  Achilles tendon  Neck/spine  Other: \_\_\_\_\_
- Muscle wasting or weakness (age of onset: \_\_\_\_\_)
  - Humeroperoneal  Scapular  Pelvic girdle  Other: \_\_\_\_\_
- Cardiac disease
  - Conduction defect/arrhythmia (describe: \_\_\_\_\_)
  - Dilated cardiomyopathy  Hypertrophic cardiomyopathy  Other: \_\_\_\_\_
- EMG findings: \_\_\_\_\_
- Other: \_\_\_\_\_

**Laboratory Findings**

- Serum CK .....  Abnormal (\_\_\_\_\_U/L)  Normal  Not performed
- Muscle histopathology .....  Abnormal (describe: \_\_\_\_\_)  Normal  Not performed
- Immunodetection .....  Abnormal (describe: \_\_\_\_\_)  Normal  Not performed

**Has the patient undergone previous germline DNA testing for muscular dystrophy?** .....  No  Yes  Unknown

If yes, describe the test(s) and results: \_\_\_\_\_  
 \_\_\_\_\_

**Is there any relevant family history of EDMD?**.....  No  Yes  Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:  
 \_\_\_\_\_  
 \_\_\_\_\_

**Has DNA testing been performed for the family member(s)?**.....  No  Yes  Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

**Check the test you intend to order.**

- 3001839 Emery-Dreifuss Muscular Dystrophy Panel, Sequencing:** Sequencing of the *EMD*, *FHL1*, and *LMNA* genes.  
Clinical sensitivity is estimated to be 36% for EDMD.
- 2001961 Familial Mutation, Targeted Sequencing:**  
Tests for a mutation previously identified in a family member.  
a copy of relative's lab result is REQUIRED.



**For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.**