

Genetic Testing for Facioscapulohumeral Muscular Dystrophy (FSHD)

There are a handful of companies in North America that offer genetic testing for FSHD. The Southern Blot test has been used the longest, while more companies are now offering whole genome optical mapping. Not all companies test for both types of FSHD, and some may test for Type 2, but separately.

Please note that the ICD-10 code for FSHD is: **G71.02**

PerkinElmer Genomics

<https://www.perkinelmergenomics.com/fshd/>

Contact:

866-354-2910

Genomics@perkinelmer.com

Testing offered:

- FSHD1 by whole genome optical mapping
- FSHD2 by full sequencing and deletion/duplication analysis of the SMCHD1 gene utilizing next-generation sequencing.
- FSHD Type 1 and Type 2 panel
- Comprehensive neuromuscular panel (including FSHD2 but NOT FSHD1)
- Comprehensive neuromuscular panel including FSHD1 testing

Requisition form: <https://www.perkinelmergenomics.com/wp-content/uploads/2022/10/68468-FSHD-req-w-IC-var.pdf>

Test	Description	CPT Codes
D8000	Type 1 testing (D4Z4 repeat size)	81404x1
D8001	Types 1 and 2 Panel	81404x1, 81479x1
D8002	Complete Neuromuscular disease panel with FSHD type 1 testing	81417x1, 81408x1, 81161x1, 81405x1, 81406x1, 81404x2

University of Iowa Diagnostic Laboratories

https://www.healthcare.uiowa.edu/path_handbook/rhandbook/test127.html

Testing offered:

•Whole genome optical mapping test for FSHD1; FSHD2 SMCHD1 sequencing; methylation testing (for FSHD1 and FSHD2). Prenatal diagnosis and 4qA/4qB allele testing. Prenatal specimens should be cultured to 100% confluency in 6 T-25 flasks and must not be frozen.

Requisition form:

https://www.healthcare.uiowa.edu/path_handbook/requisitions/FSHD1&2req.pdf?_gl=1*1v10qry*_ga*MTQzNDQzNzY2Ni4xNjk5OTg3OTcz*_ga_L7BTY7JCG8*MTY5OTk4Nzk3My4xLjAuMTY5OTk4Nzk3My42MC4wLjA.&_ga=2.232337567.1057231318.1699987973-1434437666.1699987973

Test	Description	CPT Codes
ZB5J4	FSHD1 Southern Blot test	81404
ZB5J4	FSHD2 sequencing of SMCHD1 & D4Z4 methylation assay	81479

Contact:

319-384-7212

Client Services: 866-844-2522

Andrew Costello, UI Diagnostic Laboratories Manager:
319-356-3533, Andrew-Costello@uiowa.edu

Athena Diagnostics

<https://www.athenadiagnostics.com/view-full-catalog/fshd1-southern-blot-test1>

Testing offered:

- Southern Blot test for FSHD Type 1: Test Code 405

Requisition form: <https://www.athenadiagnostics.com/ordering/supplies>

Contact:

800-394-4493, option 2

Calling from outside the U.S.: +1-508-756-2886, option 2.

Test	Description	CPT Codes
17029	FSHD1 Southern Blot test	81404(1)

Bionano Laboratories

<https://bionanolaboratories.com/fshd-test/>

801-931-6200

Clinicalsupport@bionano.com

Testing offered: optical genome mapping for FSHD Type 1 only

Other services: Genetic counseling services for patients & providers. Clinicians can reach out to the genetic counselors at: 801-931-6200.