

FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY (FSHD)

MOLECULAR DIAGNOSTIC

SAMPLING

Customer: /

Date of collection:

EDTA Whole blood (2 tubes of 5 ml) *The sample must be stored and transported at +4°C and must reach the laboratory within a maximum of 4 days after collection.*

PATIENT

LAST NAME
FIRST NAME
Maiden name
Address
ZIP Code City
Date of birth:
Mobile phone: Email Address

PRESCRIBER

Mandatory stamp

Email address:

Signature:

REQUESTED TEST

- ☐ Search for D4Z4 pathogenic contraction by optical genome mapping (OGM). *OPL code: FSHD1*
- ☐ Search for D4Z4 contraction by optical genome mapping (OGM) to +/- FSHD2 NGS panel (reflex test)
Genes: SMCHD1, DNMT3B, LRIF1, and FAT1. *OPL code: FSHD2*

CONTEXT OF REQUEST

- ☐ Investigation of a symptomatic index case: clinical suspicion of FSHD
- ☐ Investigation of a symptomatic related individual (family study) (a copy of the index case result must be provided)
- ☐ Investigation of an asymptomatic related individual (family study) (a copy of the index case result must be provided)

CLINICAL INFORMATION AND FAMILY HISTORY

Estimation of clinical severity:

- ☐ Typical form – severe to moderate ☐ Typical form – mild
☐ Atypical form – severe to moderate ☐ Atypical form – mild

Age at symptom onset: years

CPK :

Pain and/or functional complaints:.....

Facial involvement

☐ Yes

☐ No

☐ Asymmetric

☐ Symmetric

Scapular winging:

☐ Yes

☐ No

☐ Asymmetric

☐ Symmetric

Involvement of humeral muscles:

☐ Yes

☐ No

☐ Asymmetric

☐ Symmetric

Involvement of thoracic muscles:

☐ Yes

☐ No

☐ Asymmetric

☐ Symmetric

Proximal lower limb involvement:

☐ Yes

☐ No

☐ Asymmetric

☐ Symmetric

Distal lower limb involvement:

☐ Yes

☐ No

☐ Asymmetric

☐ Symmetric

Steppage:

☐ Yes

☐ No

☐ Left

☐ Right

Axial muscle involvement:

☐ Yes

☐ No Type :.....

Abdominal muscles (Beevor's sign):

☐ Present

☐ Absent

☐ No tested

Extra-muscular involvement:

☐ Cardiac involvement

☐ Respiratory involvement

☐ Ocular involvement

☐ Auditory involvement

☐ Other systemic involvement

ARBRE FAMILIAL

☐ Man

☐ Woman

☐ Individual of unknow sex

☒ Affected subject

☐ Healthy subject

Consanguinity

☐ Yes

☐ No

Death in siblings

☐ Yes

☐ No

Affected twins

☐ Yes

☐ No