



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

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	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Asymmetric	<input type="checkbox"/> Symmetric	
Scapular winging:	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Asymmetric	<input type="checkbox"/> Symmetric	
Involvement of humeral muscles:	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Asymmetric	<input type="checkbox"/> Symmetric	
Involvement of thoracic muscles:	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Asymmetric	<input type="checkbox"/> Symmetric	
Proximal lower limb involvement:	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Asymmetric	<input type="checkbox"/> Symmetric	
Distal lower limb involvement:	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Asymmetric	<input type="checkbox"/> Symmetric	
Steppage:	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> Left	<input type="checkbox"/> Right	
Axial muscle involvement:	<input type="checkbox"/> Yes	<input type="checkbox"/> No Type :.....			
Abdominal muscles (Beevor's sign):	<input type="checkbox"/> Present	<input type="checkbox"/> Absent	<input type="checkbox"/> No tested		
Extra-muscular involvement:	<input type="checkbox"/> Cardiac involvement	<input type="checkbox"/> Respiratory involvement	<input type="checkbox"/> Ocular involvement		
	<input type="checkbox"/> Auditory involvement	<input type="checkbox"/> Other systemic involvement			

ARBRE FAMILIAL

- Man
- Woman
- Individual of unknown sex
- Affected subject
- Healthy subject

Consanguinity	<input type="checkbox"/> Yes	<input type="checkbox"/> No
Death in siblings	<input type="checkbox"/> Yes	<input type="checkbox"/> No
Affected twins	<input type="checkbox"/> Yes	<input type="checkbox"/> No