

## **FSHD questions:**

### **What are the sample requirements?**

DNA prepared by any kit doesn't work for the special FSHD1-southernblots and for the FSHD1-haplotype-southernblots. Therefore, we prefer 10 ml EDTA-blood or salt-extracted very high molecular and high concentrated DNA (>50 kb and 300-600 ng/µl) from the patient. (For the FSHD2-analysis and the FSHD2-haplotype-analysis, you can send us DNA or less EDTA-blood).

The best time to send the samples is at the beginning of the week so that the sample reaches us at least by Thursday.

Please pack the sample shockproof and by room temperature. Transport costs are by your own.

Additional informations:

The new German gene diagnostics act requires us to document the patient's consent for every genetic diagnostic. Therefore, we need the patients consent for genetic analysis or a sentence like this: "The patient's consent for genetic analysis is documented in our file."

Or the whole consent:

Form for consent: [https://www.biozentrum.uni-wuerzburg.de/fileadmin/03280000/user\\_upload/5\\_Patientenversorgung/Formulare/consent-genetic-testing-engl.pdf](https://www.biozentrum.uni-wuerzburg.de/fileadmin/03280000/user_upload/5_Patientenversorgung/Formulare/consent-genetic-testing-engl.pdf)

Order forms: [https://www.biozentrum.uni-wuerzburg.de/fileadmin/03280000/user\\_upload/5\\_Patientenversorgung/Formulare/Order-single-gene-engl.pdf](https://www.biozentrum.uni-wuerzburg.de/fileadmin/03280000/user_upload/5_Patientenversorgung/Formulare/Order-single-gene-engl.pdf)

Before starting the tests, we need your written confirmation, that you will pay the costs. And for the invoice, we need your address.

### **What is the price and TAT?**

Price for FSHD1:

The FSHD1 analysis costs are about 448 €/patient.

TAT for FSHD1:

The investigation time will be about 4-6 weeks.

Price for FSHD1-haplotype-analysis:

The FSHD1-haplotype analysis costs are about 915 €/patient.

TAT for FSHD1-haplotype-analysis:

The investigation time can be up to half a year, because we are collecting samples for one protocol.

**Please keep in mind, that haplotype analysis for FSHD1 is rarely performed. As it is only recommended in sporadic and non-FSHD clinical cases or where there is no positive family history. Or**

if a prenatal test is desired at a later date. Because we perform the FSHD1 haplotype blot so infrequently, the TAT is about 6 months.

Price for FSHD2:

The FSHD2 analysis costs are about 1800 €/patient (including methylation-analysis).

TAT for FSHD2:

The investigation time will be also about 12-16 weeks.

Price for FSHD2-haplotype-analysis:

The FSHD2-haplotype-analysis costs are about 318 €/patient.

TAT for FSHD2-haplotype-analysis:

The investigation time will be about 4-6 weeks.

Do you accept samples for presymtomatic testing?

FSHD1: Yes.

FSHD2: for a confirmed variant of class 4 or 5.

Do you accept samples for prenatal testing?

FSHD1: Yes, but under following conditions: the confirmed contracted allele is smaller or equal 9 repeats and it is confirmed that the contracted repeats are on a permissive allele. A haplotype blot must have been performed.

FSHD2: We have never had this case before.

Could you briefly describe the method used?

FSHD1:

FSHD1 analysis using Southern blot EcoRI/BlnI double digestion and probe p13E-11.

FSHD1-haplotype-analysis: using Southern blot EcoRI/BlnI double digestion and HindIII digestion and probe p13E-11, probe A and probe B.

FSHD2: NGS-analysis (sequencing SMCHD1 and methylation-analysis of the D4Z4 regions)

FSHD2-haplotype-analysis: PCR (modified to Calandra et al., 2016)

Do you perform analysis for permissive alleles?

Yes, see the answers concerning the haplotype analysis.

Positive/negative analysis:

In the case of FSHD1 we report a positive report, when the contracted allele is smaller or equal 10 repeats. A negative report you will get, when we found 11 or more repeats.