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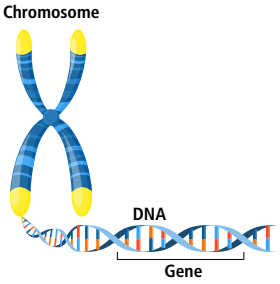
PerkinElmer GENOMICS

INTRODUCING A NEW DIAGNOSTIC APPROACH FOR FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY (FSHD)

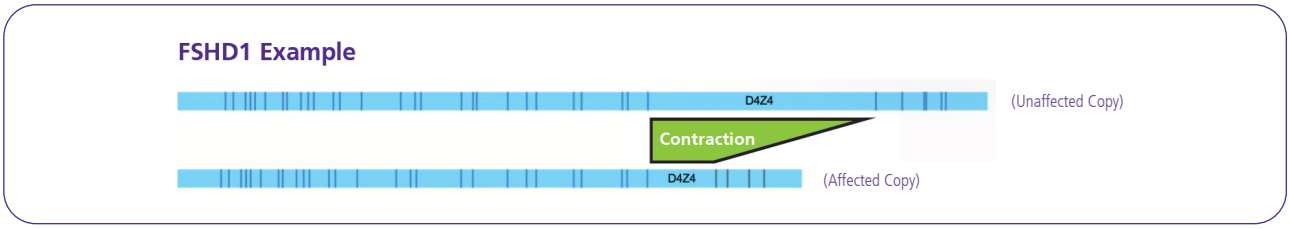


BASICS OF GENETICS

Genetics is a discipline of biology that studies susceptibility to certain conditions based on one's genetic code (DNA). Genes are specific lengths of DNA that provide instructions to our body on how to function and are packaged on worm-like structures called chromosomes. A change or a variant in a gene can sometimes result in wrong instructions for the body which can, in turn, lead to potentially developing a disorder. Variants seen in genes include misspellings in the code as well as missing or duplicated genetic materials.

GENETICS OF FSHD

FSHD is categorized into 2 types based on the underlying genetic cause with 95% of cases being type 1 (FSHD1). FSHD1 has a complex and unique genetic pattern and is caused by a contraction of a repeat unit, known as D4Z4, in the DUX4 gene located on chromosome 4. More simply put, missing codes in the DUX4 gene are responsible for developing FSHD1. FSHD2 (5%) is caused by variants in a different gene called SMCHD1.



TESTING FOR FSHD

FSHD1:
Similarities between the D4Z4 region and other regions of the DNA create challenges for traditional genetic technologies to detect D4Z4 contractions. Therefore, the availability of accurate genetic testing for FSHD1 has been limited in the past. PerkinElmer Genomics utilizes a novel approach called Whole Genome Optical Mapping to resolve that challenge.

FSHD2:
FSHD2 can be detected via an approach called sequencing. In sequencing the whole gene is read over and looked for variants in the code that could disrupt the gene function. As discussed above, FSHD2 is caused by disruptions to the SMCHD1 gene.

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TESTING MENU

PerkinElmer Genomics offers a variety of testing options for FSHD. Speak to your provider today to find the right option for you.

Test	Description	Turn Around time	Accepted Sample(s)	Billing	
				Is Insurance Accepted?	Cash-Pay
FSHD Type 1 Testing	This test includes FSHD1 testing via genome optical mapping	8-12 weeks	Whole blood	Yes, we accept insurance billing across our testing menu	$699
FSHD Type 2 Testing	This test includes FSHD2 testing via SMCHD1 gene sequencing	3 weeks	Whole blood, saliva, dried blood spot, and genomic DNA		$550
FSHD Types 1 and 2 Panel	This test both looks at FSHD1 by genome optical mapping and FSHD2 by SMCHD1 gene sequencing	8-12 weeks	Whole blood		$1,249
Complete Neuromuscular Disease Panel with FSHD Type 1 Testing	This test is an all-encompassing test looking for FSHD1, FSHD2 as well as other related neuromuscular conditions. Optical mapping method will be performed to detect FSHD1. In addition, 132 genes associated with muscular dystrophies will be sequenced, including SMCHD1 gene	8-12 weeks	Whole blood		$1,249
Comprehensive Neuromuscular Disorders Panel	This test looks at a 132 genes associated with muscular dystrophies including SMCHD1 gene. Optical mapping is not performed in this test, and it is recommended to consider FSHD Type 1 Testing to detect.	3 weeks	Whole blood, saliva, dried blood spot, and genomic DNA		$550

FREQUENTLY ASKED QUESTIONS

Who should consider getting tested for FSHD?

- You are (or your child is) suspected to have a muscular dystrophy
- You (or your child) have previously had genetic testing that did not provide a diagnosis
- Your (or your child's) current diagnosis does not seem to fully explain the symptoms
- You have a family member diagnosed with FSHD

Why is it important to get tested?

FSHD has an underlying genetic cause with potential implications for the entire family. Knowing the actual cause of the condition would help in health management of both you and your loved ones.

How to get tested at PerkinElmer Genomics?

Test must be ordered by a medical provider such as a physician, nurse practitioner, physician assistant or genetic counselor. You can contact your provider and discuss FSHD testing at PerkinElmer Genomics today.

How to choose the right testing option on the menu?

Your provider can make testing recommendations to you based on you personal and family history. Our website has information that can help in making that choice. In addition, consulting with specialists in genetics, such as a geneticist or genetic counselor, can help you decide if genetic testing is right for you and/or your family members. You can find a genetic counselor near you through the National Society of Genetic Counseling's directory: findageneticcounselor.com. We are also always available and happy to address any questions from you and your provider at Genomics@perkinelmer.com.