**Letter A: for Individuals with clinical symptoms who are seeking genetic testing**

I am writing to ask that my claim for genetic diagnosis of Facioscapulohumeral muscular dystrophy (FSHD) be approved. An accurate, **genetic** diagnosis of FSH muscular dystrophy is crucial in managing the disease, symptoms, and the quality of life for the patient.

Testing is offered through Athena Diagnostics and University of Iowa Diagnostic Laboratories.

The CPT code(s) for this testing are: 81404 (for FSHD1 Southern Blot, University of Iowa and Athena); 81404(1), 88299(1) (Molecular combing, Athena); and 81479 (for FSHD2, Methylation and SMCHD1 sequencing, University of Iowa. The appropriate ICD-10 codes are G71.0 (muscular dystrophy) or Z83 (family history of other specific disorders).

Many patients with FSHD symptoms go through years of misdiagnosis. Those years are filled with confusion, self-doubt, depression, low self-esteem, and pain, as it is hard to manage the symptoms effectively in the absence of a specific diagnosis. Misdiagnosis can lead to invasive, costly, and unnecessary diagnostic procedures (such as muscle biopsy and EMG), as well as expensive and sometimes detrimental screenings (such as echocardiograms) and treatments (steroids, orthopedic surgery).

The genetic test for FSHD diagnosis is not experimental and is medically necessary. My medical providers will be able to give me options and resources for managing the disease. Even though there are no disease-modifying treatments today, there are evidence-based interventions that are effective in managing symptoms and pain, and possibly even slowing down the progression of muscle degeneration.\* Having a definitive diagnosis will empower me to research what I can do to improve the quality of my life, make decisions around family planning, my career choices, and gave me a support group and research community dedicated to understanding this disease.

In addition, knowing my genetic status could make me eligible to participate in clinical studies and investigational drug trials that could contribute to finding treatments for FSHD.

If FSHD could be confirmed through a genetic test, I would be empowered to seek effective medical management of the symptoms while avoiding unnecessary screenings and procedures, all of which should result in reduced long-term costs for the medical care system.

Thank you for your consideration of my request.

Reference

\* Barbara H Janssen\*, Nicoline BM Voet\*, Alexander CH Geurts, Baziel GM van Engelen, Arend Heerschap. Quantitative MRI reveals decelerated fatty infiltration in muscles of active FSHD patients. *Neurology*. 2016;86(18):1700-7.

**Letter B for asymptomatic women at risk of having FSHD (because a parent or sibling has been diagnosed with FSHD).**

I am writing to ask that my claim for genetic diagnosis of Facioscapulohumeral muscular dystrophy (FSHD) be approved.

Testing is offered through Athena Diagnostics and University of Iowa Diagnostic Laboratories.

The CPT code(s) for this testing are: 81404 (for FSHD1 Southern Blot, University of Iowa and Athena); 81404(1), 88299(1) (Molecular combing, Athena); and 81479 (for FSHD2, Methylation and SMCHD1 sequencing, University of Iowa. The appropriate ICD-10 codes are G71.0 (muscular dystrophy) or Z83 (family history of other specific disorders).

The genetic test is medically important and will affect my healthcare decisions because one of my biological parents has received a diagnosis of FSHD, which puts me at a 50 percent risk of inheriting the FSHD genetic mutation. I currently do not exhibit symptoms, but recent peer-reviewed scientific research\* has documented the existence of asymptomatic individuals who have received a positive genetic diagnosis of FSHD, and that furthermore, such individuals can pass FSHD onto offspring who are symptomatic.

As a woman of child-bearing age, I need to have the FSHD genetic test and know what my genetic status is for three medical reasons:

1. To know whether I am at risk of passing FSHD onto my children;
2. To know whether pregnancy could put my health at risk\*\*;
3. To know whether I should have a periodic examination by a neuromuscular specialist to monitor for the development of muscle weakness and other symptoms.

In addition, knowing my genetic status could make me eligible to participate in clinical studies and investigational drug trials that could contribute to finding treatments for FSHD.

The results of the genetic test will influence my medical care. Pro-actively getting tested for FSHD will empower me to make informed decisions for myself and my family’s health.

Thank you for your consideration of my request.

References

\* Jones, et al. Facioscapulohumeral muscular dystrophy family studies of DUX4 expression: Evidence for disease modifiers and a quantitative model of pathogenesis. Hum. Mol. Genet. (2012) doi: 10.1093/hmg/dds284 published online: July 13, 2012.

\*\* Ciafaloni E1, Pressman EK, Loi AM, Smirnow AM, Guntrum DJ, Dilek N, Tawil R. Pregnancy and birth outcomes in women with facioscapulohumeral muscular dystrophy. Neurology. 2006 Nov 28;67(10):1887-9.

**Letter C for asymptomatic men at risk of having FSHD (because a parent or sibling has been diagnosed with FSHD).**

I am writing to ask that my claim for genetic diagnosis of Facioscapulohumeral muscular dystrophy (FSHD) be approved.

Testing is offered through Athena Diagnostics and University of Iowa Diagnostic Laboratories.

The CPT code(s) for this testing are: 81404 (for FSHD1 Southern Blot, University of Iowa and Athena); 81404(1), 88299(1) (Molecular combing, Athena); and 81479 (for FSHD2, Methylation and SMCHD1 sequencing, University of Iowa. The appropriate ICD-10 codes are G71.0 (muscular dystrophy) or Z83 (family history of other specific disorders).

The genetic test is medically important and will affect my healthcare decisions because one of my biological parents has received a [genetic diagnosis / a clinical diagnosis] of FSHD, which puts me at a 50 percent risk of inheriting the FSHD genetic mutation. I currently do not exhibit symptoms, but recent peer-reviewed scientific research\* has documented the existence of asymptomatic individuals who have received a positive genetic diagnosis of FSHD, and that furthermore, such individuals can pass FSHD onto offspring who are symptomatic.

The FSHD genetic test is medically necessary for me because:

1. I need to know whether I am at risk of passing FSHD onto my children;
2. I need to know whether I should have a periodic examination by a neuromuscular specialist to monitor for the development of muscle weakness and other symptoms.

In addition, knowing my genetic status could make me eligible to participate in clinical studies and investigational drug trials that could contribute to finding treatments for FSHD.

The results of the genetic test will influence my medical care and family planning choices. Pro-actively getting tested for FSHD will empower me to make informed decisions for my health and my family’s future health and well-being.

Thank you for your consideration of my request.

Reference

\* Jones, et al. Facioscapulohumeral muscular dystrophy family studies of DUX4 expression: Evidence for disease modifiers and a quantitative model of pathogenesis. Hum. Mol. Genet. (2012) doi: 10.1093/hmg/dds284 published online: July 13, 2012.

**Letter D for asymptomatic parent whose child has been diagnosed with FSHD**

I am writing to ask that my claim for genetic diagnosis of Facioscapulohumeral muscular dystrophy (FSHD) be approved.

Testing is offered through Athena Diagnostics and University of Iowa Diagnostic Laboratories.

The CPT code(s) for this testing are: 81404 (for FSHD1 Southern Blot, University of Iowa and Athena); 81404(1), 88299(1) (Molecular combing, Athena); and 81479 (for FSHD2, Methylation and SMCHD1 sequencing, University of Iowa. The appropriate ICD-10 codes are G71.0 (muscular dystrophy) or Z83 (family history of other specific disorders).

The genetic test is medically important and will affect my family’s healthcare decisions. One of my biological children has received a genetic diagnosis of FSHD. Because the disease is hereditary in over 70 percent of cases, it is likely that I too have the FSHD genetic mutation. I currently do not exhibit symptoms, but recent peer-reviewed scientific research\* has documented the existence of asymptomatic individuals who have received a positive genetic diagnosis of FSHD, and that furthermore, such individuals have a 50-50 chance of passing FSHD onto each child, putting them at risk of developing the disease.

The FSHD genetic test is medically necessary for me and my family because:

1. I need to know whether I am at risk of passing FSHD onto my other children;
2. If my children are at risk of having inherited the FSHD mutation, as adults they should be given the option to be tested themselves so that they can make informed healthcare decisions;
3. I need to know whether I should have a periodic examination by a neuromuscular specialist to monitor for the development of muscle weakness and other symptoms.

In addition, knowing my genetic status could make me eligible to participate in clinical studies and investigational drug trials that could contribute to finding treatments for FSHD.

The results of the genetic test will influence important medical care decision for me and my family.

Thank you for your consideration of my request.

Reference

Jones, et al. Facioscapulohumeral muscular dystrophy family studies of DUX4 expression: Evidence for disease modifiers and a quantitative model of pathogenesis. Hum. Mol. Genet. (2012) doi: 10.1093/hmg/dds284 published online: July 13, 2012.