Testing and diagnosis for DM

Making an initial diagnosis starts with a complete family history and physical examination. A person will also undergo a battery of medical tests, depending on the symptoms he or she is having. A key element of the evaluation is electromyography (EMG). This procedure detects the presence of myotonia in a high proportion of people with DM1 or DM2. When test results point strongly toward a diagnosis of myotonic dystrophy, the disorder can be confirmed by genetic testing.

The genetic test requires a blood sample from the patient. The DNA is extracted from the blood sample and analyzed to see if that person has the mutation that causes myotonic dystrophy. Prenatal testing, where the DNA of the fetus is checked for the presence of the myotonic dystrophy mutation, is also available.

Diagnosis of myotonic dystrophy is not difficult once the disorder is suspected. However, the path to a correct diagnosis of myotonic dystrophy can be long and complex, and delays in diagnosis are very common. It can typically take over 6 years to reach a diagnosis of DM1 and up to 11 years to confirm DM2.

Genetic testing

Genetic testing (also referred to as DNA testing) is a definitive diagnostic of whether or not a person has DM. DNA, the genetic material in the nucleus of cells, is isolated from a sample of blood or other tissue, and then analyzed to determine whether or not a specific mutation is present.

Genetic testing is available for DM1 using standard DNA diagnostic protocols (PCR and southern blot) to confirm the presence of DM.

Genetic testing is also available for the diagnosis of DM2 using standard DNA diagnostic protocols. However, in some cases the repeat expansion for DM2 may be too large for PCR testing. In those instances, southern blot techniques are used for diagnosis.

Reasons to consider genetic testing

Genetic testing can be beneficial in the following situations:

- A confirmed diagnosis can eliminate the need for additional medical tests and reduce anxiety about the cause of symptoms.
- People with DM should be educated about the dangers of anesthesia and alert their doctors if they should need surgery.
- Couples can make family planning decisions based on their genetic risk.
- Mothers with DM1 can have special monitoring during pregnancy and prepare for risks involved for a child born with congenital DM.

Obstacles to getting a diagnosis

Since the symptoms of DM often mimic more common diseases, many potential causes need to be ruled out through medical testing. Medical professionals are often unfamiliar with DM since they see these cases so infrequently. Selection of the appropriate genetic test may not be obvious since there are more than thirty genetically distinct forms of muscular dystrophy. In the case of myotonic dystrophy, diagnosis is complicated even further by the variability of the disease. Some of the confounding issues include:

- Myotonic dystrophy can take multiple forms that affect a broad spectrum of systems. Individuals may visit several different specialists for disparate symptoms, such as an ophthalmologist for blurred vision, a gastroenterologist for stomach pain, and a cardiologist for an abnormal heartbeat. These individual physicians may not be aware of their patient's full range of problems and therefore may not be able to put the pieces together for an accurate diagnosis.
- The severity of symptoms can vary greatly, even within the same family. Quite often individuals go to their primary care physician with a variety of complaints, most so general that the doctor never suspects any serious problem. As a result, a correct diagnosis may be delayed until the disease has progressed significantly.