How is muscular dystrophy diagnosed?

En Español

The first step in diagnosing muscular dystrophy (MD) is a visit with a health care provider for a physical exam. The health care provider will ask a series of questions about the patient's family history and medical history, including any problems affecting the muscles that the patient may be experiencing.

The health care provider may order tests to determine whether the problems are a result of MD and, if so, what form of this disorder. The tests may also rule out other problems that could cause muscle weakness, such as surgery, toxic exposure, medications, or other muscle diseases.[1] These tests may include:[2]

- **Blood tests** to measure levels of serum creatine kinase, an enzyme that is released into the bloodstream when muscle fibers are deteriorating, and serum aldolase, an enzyme that helps break down sugars into energy. Elevated levels of either of these enzymes can signal muscle weakness and indicate a need for additional testing.
- **Muscle biopsies**, which involve the removal of muscle tissue using a biopsy needle. The tissue is then examined under a microscope to provide information on the amount and level of the genes that may cause MD. Patients diagnosed by muscle biopsy typically require genetic testing as well to determine any mutations in their genes.[3]
- **Genetic testing** to evaluate missing or repeated mutations in the dystrophin gene. A lack of the dystrophin gene can lead to a diagnosis of Duchenne or Becker MD. The test is important not only to confirm the MD diagnosis in males but also to determine whether women with a family history of Duchenne or Becker MD may be carriers.[4]
- **Neurological tests** to rule out other nervous system disorders, to identify patterns of muscle weakness and wasting, to test reflexes and coordination, and to detect contractions.
- **Heart testing**, such as an electrocardiogram (ECG), to measure the rate and frequency of heartbeats. Some forms of MD cause heart problems, such as an irregular heartbeat.[5]
- **Exercise assessments** to evaluate the patient's level of strength and respiratory function and to detect any increased rates of certain chemicals such as nitric oxide following exercise.
- **Imaging tests** such as magnetic resonance imaging (MRI) and ultrasound imaging. These painless tests use radio waves (MRI) and sound waves (ultrasound) to obtain pictures of the inside of the body and to examine muscle quality and bulk as well as the fatty replacement of muscle tissue.

Read more at https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Hope-Through-Research/Muscular-Dystrophy-Hope-Through-Research#3171_7.