

Slide presentation at the 7th International Myotonic Dystrophy Conference - September 2009

Spectrum of disease manifestations of juvenile myotonic dystrophy type 1 (JDM) patients

Background

The full spectrum of the clinical manifestations of juvenile myotonic dystrophy (JDM), or childhood-onset myotonic dystrophy, is not entirely understood. Little information is available on the symptoms, progression of the disease over time, and other characteristics of JDM and how these characteristics compare or contrast with patients having congenital (from birth) and adult-onset forms of DM1.

We investigated the impact of disease in congenital DM (CDM) and JDM by analyzing patient reported data and reviewing patient medical records of members of the National Registry. JDM patients were defined as having an onset of their symptoms before reaching 11 years of age and having an uneventful prenatal and neonatal history. The JDM patients had no severe myotonic dystrophy symptoms within the first year of life. CDM patients were defined as having symptoms within the first 4 weeks of life.

Results

The National Registry has enrolled 23 JDM members and 33 CDM members as of September 2009. JDM and CDM members represent 3.5% and 5.1% of all DM1 members enrolled in the Registry. JDM patients have an average age of 5.9 years at symptom onset. Learning difficulty is the most frequently reported first symptom (26.1%). Myotonia, developmental delay, and weakness are also common first symptoms.

Compared to JDM patients, CDM patients in the Registry have greater use of physical, occupational and speech therapies as well as assistive devices. CDM patients also have a history of more chronic infection, constipation, and pneumonia. JDM patients report a greater spectrum of diverse symptoms at their initial clinical presentation. They report more pacemaker use, and more acid reflux and asthma. JDM patients also have a greater use of psychological counseling and more psychological disorders. Attention deficit/hyperactivity disorder (ADHD) is the most common psychological disorder for both groups.

Conclusion

The distinct variations between CDM and JDM patients indicate a need for further research to better characterize these DM1 subtypes. For example, CDM patients report fewer psychological problems compared to JDM. One explanation for this may be that CDM patients are more severely affected and have more immediate care concerns compared to JDM and these concerns, both psychological and physical, are being addressed early by physical, occupational, and speech therapy. JDM patients are also significantly older at enrollment into the Registry and are likely to have more psychological concerns that develop later in childhood. Many opportunities exist to study the pathophysiology of cognitive impairment in DM1, and whether the mechanisms in JDM compare or contrast to CDM and adult-onset DM1. Other important opportunities for studies involve further assessment of disease manifestations and progression, quality of life, and symptom management in JDM.