

## **Slide presentation at the International Myotonic Dystrophy Conference 6- September 2007**

### **Gastrointestinal (GI) symptoms in myotonic dystrophy type 1 (DM1) patients enrolled in the NIH Registry**

#### **Background**

Gastrointestinal (GI) problems are a frequent and serious complaint in DM1 patients. The cause of GI disturbances in DM1 remains unclear and limited information is available about the overall prevalence of symptoms and most frequently used treatments.

We reviewed information provided by members of the Registry in order to examine the prevalence of GI symptoms and the types of medications that are used. We also investigated if other characteristics (age of symptom onset, weight, genetic repeat size) were linked to GI symptoms.

#### **Results**

The Registry had enrolled 479 members diagnosed with DM1 in September 2007. Of these enrolled members, 361 patients (75%) reported experiencing GI symptoms. The most common GI-related complaints were difficulty swallowing, gastro-esophageal reflux disease (GERD), and constipation. Patients with GI symptoms were taking a total of 131 medications for GI related symptoms. The most common type of medication used were acid blockers, followed by medications to control constipation, and prokinetics (medications that stimulate contractions of the stomach muscles). The presence of GI symptoms was not related to the age at which DM1 symptoms appeared or to a patient's CTG repeat size. GI symptoms were more common in patients that were overweight and had other symptoms of DM1 for a long period of time.

#### **Conclusion**

The large number of patients experiencing GI issues indicates a need for improvements in symptom management, including studies of the effectiveness of different pharmacological, nutritional, and other therapies. Further investigations into the underlying cause(s) of GI dysfunction in DM1 are needed.

#### **Future research of Registry patients**

Updated results from this investigation will be presented at the upcoming Annual Meeting of the American Neurological Association, in September 2008. This report will include new information about GI symptoms in patients with myotonic dystrophy type 2 (DM2) and updated information on GI symptoms in DM1. This information will be available on the Registry website in the near future.