Beyond Myotonia

Having worked with hundreds of myotonia patients over the last twenty years, I often see presentation of symptoms that seem to go beyond what would be typical for myotonia alone. Here is a list of observed co-existing conditions that are probably coincidental but have occurred in myotonia patients and further affected muscle function.

Ehlers-Danlos Syndrome
The hypermobility type of EDS is probably the most common co-existing condition that we see. When the joints are already unstable from the collagen defect (the exact genetic cause with Type III is not known yet), myotonia can cause more frequent dislocations and subluxations. This results in greater pain and wear and tear on the joints. Many develop fibromyalgia.

Glycogen Storage Disease
The two types I have seen in patients is McArdle Disease and Pompe's Disease. When a person has severe cramping with exercise or exercise intolerance (and paramyotonia has been excluded) then I suggest checking for glycogen storage issues. This is especially important if there is abnormal weakness or darkened urine suggesting rhabdomyolysis.

Mitochondrial Disease
While the more severe forms of these diseases are evident in infancy and childhood, there are milder forms that can persist throughout life. When someone with myotonia has unusual weakness and exercise intolerance, it can be from a mild complex deficiency. Just supplementing carnitine and using MCTs can often make a significant difference in muscle damage and shorten recovery time. With myotonia we already tend toward lactic acidosis and while doctors often test CK levels, they don't usually look for acidosis. If there is any type of cardiomyopathy in someone with myotonia, mitochondrial disease should be investigated.

Magnesium Deficiency
While this isn't usually a genetic condition, it is very common and contributes to excessive pain and cramping with myotonia. The form of magnesium used can make some difference, magnesium chloride and magnesium glycinate being more effective without causing as much of a laxative effect. Sodium intake needs to be adequate when supplementing.

Hypothyroidism
This is another very common acquired condition that can have a dramatic effect on the severity of myotonia. My endocrinologist who was familiar with the correlation has a guideline of .5 to 1.5 for TSH levels. Many doctors refuse to prescribe thyroid hormone replacement until levels are above 6. For someone with myotonia, levels above 3 can be enough to start causing a noticeable worsening of symptoms.

Tetany
Occasionally tetany is seen along with myotonia. It can be hard to distinguish, but if someone has muscle spasms and "charley horses" I suggest checking for this. Tests would include serum calcium, parathyroid function and vitamin D status. Sometimes supplementing with vitamin D will improve myotonia.

Medication-Induced Myotonia
Many medications will worsen myotonia, statin drugs and quinolone antibiotics being the most common.