

Specific FAQ's – answers from MDF's Scientific Advisory Board

- 1. If two siblings have the disease, can one expect them over time to manifest similar organ system involvement?**

Not always. Their genetic background is different although many genes are shared. The genomic background is likely to play an important role in organ-specific phenotype expression.

- 2. If you know you have the disease, are there ways to have children who are not affected? Are there any additional risks for an affected woman during pregnancy? Are there any precautions she should take if she becomes pregnant?**

PGD and prenatal testing will be useful, given the couple accept the ethical implications. Risks for the pregnant women consist of polyhydramnios, placenta previa, miscarriage, preterm birth, stillbirth, and complications in labor and delivery including prolonged labor, anesthesia risks and postpartum hemorrhage. Psychological risks, especially after having a baby with the congenital form, should be considered.

- 3. Is there always an expansion at every generation? Is it larger with maternal transmissions? Or is there an identical distribution between men and women?**

Not always. About 6% of paternal transmissions result in contraction of the repeat in the offspring. Expansion is more prominent with paternal transmissions when the repeat is small (37-100) while it is much larger with maternal transmissions when the repeat is over a few hundred.

- 4. Is there new information on the impact on the brain from DM and how is the executive function affected, especially in childhood onset?**

There is only one paper that introduced a new concept that assesses the progressive nature of cognitive dysfunction in some childhood-onset patients (Echenne, et al. 2007 Eur J Paediatr Neurol). Brain involvement is an aspect of DM that several research centers, including the University of Minnesota, are keenly interested in; we will post their findings when published.

- 5. Regarding anesthetic risks, what specifically should one tell an anesthesiologist before surgery?**

Perioperative complication is increased in patients with myotonic dystrophy. All drugs, including sedatives, induction drugs, anesthetics, neuromuscular junction blockers and opiates must be carefully chosen, and doses must be carefully determined. In particular, anticholinesterases (e.g., neostigmine), depolarizing neuromuscular blocking agents (e.g., suxamethonium) and inhalational anesthetics should be avoided. Cardiac problems should be alerted to the anesthesiologist. The anesthesiologist should also be aware that hyperkalemia, hypothermia and shivering and mechanical or electrical muscle stimulation can cause myotonia which may interfere with the surgery. Perioperative aspiration is a risk due to bulbar weakness.

6. **If one develops serious cardiac problems --- a very rapid or very slow heart beat, or arrhythmia (irregular heartbeat) --- what sort of device is generally implanted in one's chest? How should a DM patient be followed from a cardiac standpoint -i.e. EKG's, echos, etc.**

The patient should have an EKG, echo, and electrophysiological (EP) studies, depending on the nature of arrhythmia to determine the need for a pacemaker. If a pacemaker needs to be implanted, a device with pacemaker/defibrillator capability may be preferable.

7. **Many patients have many problems with diarrhea and constipation. Are these related to the following: a) digestion, b) type of food eaten, or c) muscles not working properly? How can these problems be treated?**

Most problems are due to intestinal motility. Selection of foods is important. Appropriate amount of fiber supplements may be useful, although overuse may produce impaction if the patient becomes constipated. Stool softener and non-irritant laxatives are useful. Cisapride (Propulsid) and other prokinetic drugs should be avoided because of cardiotoxicity.

8. **What does one do when swallowing becomes a problem? What emergencies should be followed? Does chewing food a lot help food go down easily? Does drinking lots of liquids with a meal help? Any particular type of liquid?**

Swallowing problems come from both oropharyngeal muscle weakness and abnormal motility of the esophagus. The patient should be evaluated by a speech pathologist and a gastroenterologist with fibroscopic and manometric testing. If patients are not able to do these studies, the modified barium swallow test should be done to assess the risk for aspiration. The speech therapist should be able to give useful advice to alleviate the problem. If the problem imposes high aspiration risks, G tube insertion should be considered. Chewing, drinking fluids, and pureed foods may help. If aspiration occurs, or may have occurred and now fever starts, go to an emergency center for treatment.

9. **What can be done to help patients who need ventilators to establish a beneficial level of support?**

For those who have a tracheotomy, the right answer may be "as needed" if the patient can get off the ventilator intermittently. Keeping the patient comfortable and able to communicate is important.

10. **Recognizing that exercise does not prevent the progression of muscle weakness in DM, are there exercise regimens that are recommended to try and maintain what muscle strength is present?**

Overdoing is counter-productive. Low intensity aerobic training may be useful.

11. **How reliable is pre-implantation DNA analysis of embryos and is there information on rate of successful pregnancy when the woman does not have DM?**

It is increasingly available. Kakourou et al. "Preimplantation genetic diagnosis for myotonic dystrophy type 1 in the UK." *Neuromuscul Disord* [Epub ahead of print] is the newest paper available.

12. Is there a medication to help with daytime sleepiness? What is the relationship between sleep apnea and DM?

Modafinil is the choice although it is expensive. Sleep apnea contributes to the daytime somnolence but patients often continue to have daytime sleepiness after CPAP.