

Supplemental Online Content

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eTable. Management of Individuals With Monotonic Dystrophy Type 1

This supplemental material has been provided by the authors to give readers additional information about their work.

eTable. Management of Individuals With Myotonic Dystrophy Type 1

System	Symptoms and Clinical Presentations	Management
Surgery and anesthesia	<ul style="list-style-type: none"> Hypersensitivity to sedatives/analgesics Paradoxical reaction to muscle-depolarizing agents Cardiac conduction defects/arrhythmias Ventilatory insufficiency or poor airway protection Gastrointestinal dysmotility 	<ul style="list-style-type: none"> - Patient and neuromuscular clinician should meet prior to surgery to discuss anesthesia and surgery risks - Minimize complications during surgery with airway care, choice of induction agents, local anesthesia, and neuromuscular blockade. - Consider prophylactic defibrillator/pacemaker - Monitor for aspiration and gastrointestinal pseudo-obstructions in the post-operative period
Respiratory	<ul style="list-style-type: none"> Ineffective cough Recurrent pulmonary infections FVC<50% of predicted or maximum inspiratory pressure (MIP)<60 	<ul style="list-style-type: none"> - Respiratory function testing at baseline and annually thereafter (FVC, FEV1, MIP, maximum expiratory pressure (MEP), nocturnal oximetry, and cough peak expiratory flow) - Patients should stay up to date on recommended vaccinations
Cardiovascular	<ul style="list-style-type: none"> Sinus bradycardia Progressive heart block Atrial fibrillation or flutter Ventricular tachycardia 	<ul style="list-style-type: none"> - ECG at diagnosis; annually thereafter - Treat with guidelines for ICD/pacemaker placement - Referral to cardiologist is warranted for any cardiac abnormalities <p style="text-align: center;">**Of note, heart failure is not considered to be a defining symptom</p>

System	Symptoms and Clinical Presentations	Management
Gynecology and obstetrics	<p>Gynecological:</p> <ul style="list-style-type: none"> - Painful or irregular menses - Infertility <p>Obstetrics:</p> <ul style="list-style-type: none"> - Accelerated myotonic dystrophy type 1 progression - Preeclampsia - Prolonged labor - Spontaneous abortion - Placenta previa - Peripartum hemorrhage 	<ul style="list-style-type: none"> - Carefully evaluate patients' reproductive history, myotonic dystrophy-related personal history, and myotonic dystrophy family history - Referral to a high-risk obstetrician (maternal-fetal medicine specialist) for prenatal care and delivery - Cesarean delivery should be carefully considered - anesthetics or sedating anesthetic drugs should be used with caution (see surgery, anesthesia, and pain section) - Family planning is recommended; diagnostic testing (preimplantation genetic diagnosis, amniocentesis, or chorionic villus sampling) paired with counseling can determine if the fetus is affected
Musculoskeletal	<p>Skeletal muscle weakness</p> <ul style="list-style-type: none"> - Bulbar/ocular muscles - Finger flexors and ankle dorsiflexors - Abdominal or diaphragm 	<ul style="list-style-type: none"> - Annual evaluation to identify swallowing or speech difficulties, difficulty with mobility/balance (risk of falls) - Evaluation by physical or occupational therapist for assistance with orthotics, assistive devices, or activities of daily living - Moderate/low-intensity aerobic and resistance exercise
	Myotonia	- Mexiletine as symptomatic treatment
Ocular	Cataracts	- Annual eye exam; slit-lamp exam to look for multicolored, iridescent, punctate opacities located in posterior lens capsule
Gastrointestinal	<p>Dysphagia, dysarthria, aspiration</p> <p>Gastroesophageal reflux</p> <p>Irritable bowel symptoms</p>	<ul style="list-style-type: none"> - Referral to a speech therapist - Modified barium swallow or barium upper GI radiographic evaluation - Screening with a glucose breath test for bacterial overgrowth syndrome

System	Symptoms and Clinical Presentations	Management
Central nervous	Excessive daytime sleepiness Sleep apnea	- Respiratory function testing and sleep study. - Stimulant therapy with modafinil or armodafinil
	Intellectual disability Impaired attention Mood/anxiety disorders Abnormal visual-spatial abilities Delusions or psychotic features Apathy	- Annual neuropsychological evaluation is recommended with insight from patients' family or partner - Psychostimulants can be warranted if apathy is associated with an impairing level of fatigue or excessive daytime sleepiness - Referral to a mental health care professional when evaluation indicates psychiatric/behavioral abnormalities
Endocrine and metabolic	Hyperlipidemia Diabetes Mellitus (T1DM, T2DM) Thyroid Dysfunction Liver Enzyme Elevation Gonadal Insufficiency Erectile Dysfunction	- Test for endocrine/metabolic perturbations at baseline and annually thereafter (blood glucose, serum lipids, thyroid levels, liver enzymes and bilirubin) a. Chronic liver enzyme elevation is not uncommon and alone does not warrant a liver biopsy - Impact of statins is unknown, clinicians should monitor for muscle side effects - Consider erectile dysfunction medications but be cautious of hypotension
Tumors	Increased risk of cancer	- Regular screenings for patient's age and sex - Encouraged to closely monitor for the following cancer types: a. Colon, breast, testicular, cervical, skin (pilomatrixomas) ^a

^aSource: D'Ambrosio ES, Gonzalez-Perez P. Cancer and myotonic dystrophy. *J Clin Med.* 2023;12(5):1939. Medline:36902726 doi:10.3390/jcm12051939