Myotonic Dystrophy (DM): Overview of Screening and Management

Background

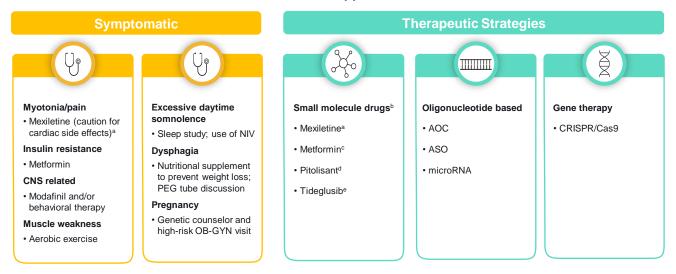
- As a multisystem disease, DM can present with a variety of symptoms best evaluated by multidisciplinary screening. Subtypes (DM1, DM2) are confirmed via genetic testing, and additional subtypes are defined based on symptom onset and severity
- Current treatment is based on a multidisciplinary approach (including small molecule therapies) to reduce symptoms. Additional therapeutic strategies are being evaluated through preclinical and clinical research

Screening: Multidisciplinary Approach^{1,2}



AFOs, ankle-foot orthoses; PEG, percutaneous endoscopic gastrostomy; PFT, pulmonary function test; PWCs, power wheelchairs.

Treatment Approach^{3–8}



^aUnder investigation for efficacy and safety in a phase 3 trial for myotonia in patients with DM1 and DM2 (NCT04700046) by reducing DMPK mRNA levels.^{4 b}These repurposed drugs, which were initially developed to treat a different condition, are being investigated in clinical trials for their possible therapeutic use in DM1, and not yet approved for this condition. ^CUnder phase 3 investigation for efficacy and safety, to improve muscle function by correcting splicing defects, in patients with DM1 (NCT03692312). ^dUnder phase 2 investigation for efficacy and safety, for excessive daytime sleepiness in patients with DM1 (NCT03692312) by reducing expression of DMPK RNA.⁴

AOC, antibody oligonucleotide conjugate; ASO, antisense oligonucleotide; CNS, central nervous system; CRISPR, clustered regularly interspaced short palindromic repeats; DM, myotonic dystrophy; DMPK, dystrophia myotonica protein kinase; NIV, noninvasive ventilation; OB-GYN, obstetrician-gynecologist; PEG, percutaneous endoscopic gastrostomy.

Additional observational and biomarker studies are ongoing to help inform clinical phenotype/genotype, less invasive assessment methods (vs muscle biopsy), and clinical endpoints⁹

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