

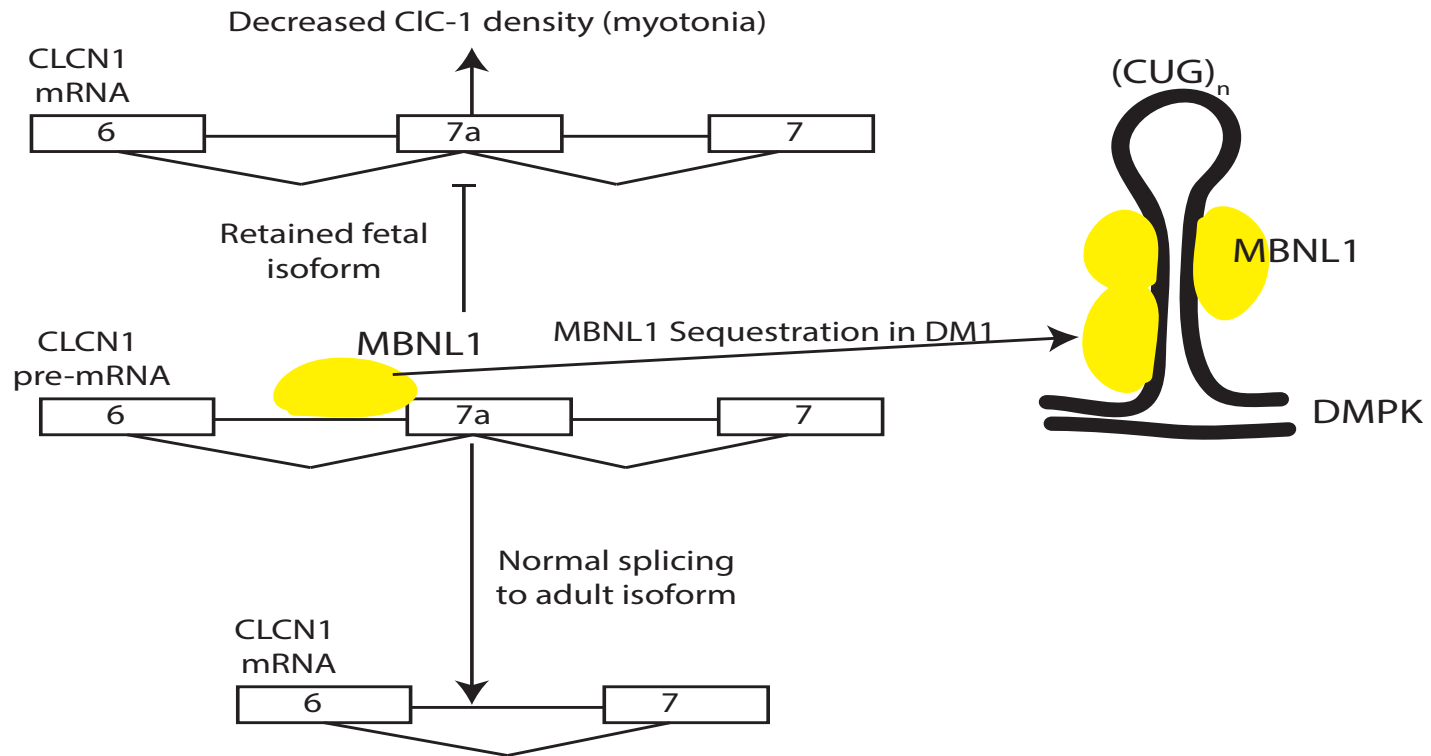
# Overview of Myotonic Dystrophy Symptoms and Pathology

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# Myotonic Dystrophy Type-1 Pathogenesis

- Autosomal dominant disorder
- CTG repeat expansion in DMPK on chromosome 19q13.3 leads to MBNL sequestration



# Core Symptoms

- Distal muscle weakness and wasting
  - Progressive hand and foot weakness
- Myotonia (delayed muscle relaxation)
- Early onset cataracts

# A Multisystemic Disorder

- Cardiac arrhythmias
  - Increase with age and weakness
  - Progressive heart block, prolonged QRS and PR interval
  - Often require a pacemaker
- Gastrointestinal symptoms
  - Abdominal pain, constipation, diarrhea, fecal incontinence
  - Dysphagia
- Endocrine abnormalities
  - Glucose intolerance, thyroid dysfunction, testosterone deficiency



# A Multisystemic Disorder

- Cognition impaired
  - Executive function and visual spatial processing deficits
  - Global cognitive dysfunction
  - Avoidant personality disorder
- Prominent daytime sleepiness and fatigue
  - Number one symptomatic issue
  - Combination of OSA and central hypoventilation
- Respiratory
  - Respiratory failure
  - Anesthesia risks
- Increased risk of neoplasms
  - Increased risk of endometrial, brain, ovarian, and colon

# Epidemiology of Myotonic Dystrophy

- Prevalence 1:8000 (most common adult muscular dystrophy)
- Average age of death 53
- Cause of Death (n=20)
  - Cardiac arrhythmias or heart failure (6)
  - Infection (4)
  - Neoplasm (2)
  - Stroke (2)
  - Unspecified (6)

# Relative Risk of Comorbidities

Co-morbidity	Relative Risk	P-value	Mean Age at Diagnosis of Comorbidity (years) (SD; range)
Cardiac Conduction Disorder	60.2 (29.9, 108.6)	<0.0001	39.6 (11.3; 18-48)
Obstructive Sleep Apnea	9.8 (5.6, 15.8)	<0.0001	45.4 (13.4; 22-74)
Central Sleep Apnea	66.1 (11.7, 208.1)	<0.0001	*
Intellectual Disabilities	9.0 (1.6, 28.4)	0.02	*
Hypothyroidism	2.6 (1.1, 5.1)	0.03	51.5 (21.8; 27-79)
Any Cataract	6.23 (4.08, 9.15)	<0.0001	41.3 (12.4; 20-59)

\* -- indicates that the exact number is below 5 and cannot be reported due to data confidentiality

# Routine Care in DM

Organ system	Screening test	Time
Heart	Routine ECG	Once yearly or symptomatic
Respiratory	Supine FVC	At diagnosis and symptomatic
Sleep (CNS)	Polysomnogram	Symptomatic
Eye	Slit lamp exam	Once yearly or symptomatic
Ear	Audiometry	symptomatic
Endocrine	HbA1c, TSH, lipid panel	At diagnosis and once yearly
Malignancy	Routine surveillance for age + consider CNS tumors, endometrial tumors	Symptomatic