Commonalities between DM1 and DM2

	DM1	DM2
Core Features*		
Myotonia	++	(+) to +, on EMG
Muscle Weakness	++	(+) to ++
Cataracts	++	- to ++
Localization of Muscle Weakness		
Facial Weakness, Jaw Muscles	++	- to +
Distal Limb Muscle Weakness	++	- to +
Proximal Limb Muscle Weakness	(+)	+ to ++
Sternocleidomastoid Muscle	++	+ to ++
Muscle Symptoms		
Muscle/Joint Pain and Stiffness	-	- to ++
Muscle Strength Variations	-	- to +
Muscle atrophy	++, distal	- to +
Muscle Cramps	- to (+)	- to +
Calf Hypertrophy	-	- to ++
Muscle Biopsy		
Fiber Atrophy	- to +, type-1 fibers	+ to ++, type-2 fibers
Cardiac Arrhythmias	++	- to ++
Elevated Serum CK Levels	(+) to ++	(+) to ++
GGTase Elevation	+	- to +
Hypoimmunoglobulinemia IgG	+	- to +
Hyperhydrosis	-	- to +
Brain		
Tremors	-	- to ++
Late Change in Mental State	++	- to (+)
Hypersomnia	+	- to (+)
Mental Retardation	+, congenital form	-
Insulin Resistance/Glucose Intolerance/Diabetes	+	- to (+)
Male Hypogonadism	+	- to +
Frontal Baldness	++	- to (+)
Genetics		
Inheritance	AD	AD
Anticipation	++	- to (+)
Locus	*DMPK*	*ZNF9*
Chromosome	19q13.3	3q21.3
Expansion Mutation	(CTG)n	(CCTG)n
Congenital Form	+	-

*+, present; ++, pronounced; (+), variably present; -, absent. AD, autosomal dominant.

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Differences in Clinical Manifestations between DM1 and DM2

	DM1	DM2
Genetics		
Inheritance	autosomal dominant	autosomal dominant
Anticipation	pronounced	exceptional
Congenital Form	yes	no
Chromosome	19q13.3	3q21.3
Locus	*DMPK*	*ZNF9*
Expansion Mutation	(CTG)n	(CCTG)n
Location of Expansion	3' UTR	Intron 1
Core Features		
Clinical Myotonia	evident in adult onset	present in <50%
EMG Myotonia	generally present	absent and variable in many
Muscle Weakness	disabling by age 50	onset as late as age 60-70
Cataracts	generally present	present in minority
Localization of Muscle Weakness		
Facial Weakness, Jaw Muscles	generally present	usually absent
Bulbar Weakness - Dysphagia	generally present later in life	not present
Respiratory Muscles	generally present later in life	exceptional cases
Distal Limb Muscle Weakness	generally prominent	flexor digitorum profundus on testing, but only in some
Proximal Limb Muscle Weakness	may be absent	main disability in most patients, late
Sternocleidomastoid Weakness	generally prominent	prominent in few
Muscle Symptoms		
Myalgic Pain	absent or mild	most disabling symptom in many
Muscle Strength Variations	no variations	can be considerable
Visible Muscle Atrophy	face, temporal, distal hands and legs	usually absent
Calf Hypertrophy	absent	present in ≥50%
Muscle Biopsy		
Fiber Atrophy	smallness of type-1 fibers	highly atrophic type-2 fibers
Nuclear clump fibers	in end-stage only	scattered early before weakness
Sarcoplasmic Masses	very frequent in distal muscles	extremely rare
Ring Fibers	frequent	may occur
Internal Nuclei	massive in distal muscle	variable and mainly in type-2 fibers
Cardiac Arrhythmias	generally present	highly variable: absent to severe
Brain		
Tremors	absent	prominent in many
Behavioral Changes	early in most	not apparent
Hypersomnia	prominent	infrequent
Cognitive Decline	prominent	not apparent
Manifest Diabetes	frequent	infrequent
Frontal Balding in Males	generally present	exceptionally
Other Features		
Childhood-onset CNS-problems	frequent	absent
Increased frequency of co-segregating heterozygous recessive *CLCN1* mutations	absent	present
Incapacity (Work and ADL)	always >30-35	rarely <60 unless severe pains
Life Expectancy	reduced	normal range

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