The Humanistic Burden of Myotonic Dystrophy Type 1: A Literature Review

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Complete Study Listing

Table 1: Study Listing

Author, Year	Country	Sample Size	Study Type	Type of Humanistic Burden Reported	Key Instruments Used
Baldanzi 2016[1]	Italy	65	Observational	Patient QoL and Mental Health	INQoL, AES, BDI, TMT, MIRS
Balloy 2020[2]	France	32	Observational	Patient QoL	GIQLI
Beauchesne 2021[3]	Canada	72	Observational	Patient QoL	EQ-5D
Callus 2018[4]	Italy	31	Observational	Patient Mental Health	SCL-90-R, MMSE, FAB, ENB-2
Endo 2019[5]	Japan	51	Observational	Patient QoL and Mental Health	SF-36 (v2), CES-D, ESS
Fujino 2018[6]	Japan	60	Observational	Patient QoL and Mental Health	MDQoL, CAT, TMT, PHQ-9, ESS, MFI
Gallais 2015[7]	France	38	Observational	Patient QoL and Mental Health	LARS, Walton Functional Scale , MMSE, KFSS, MINI
Hagerman 2019[8]	US + Canada	1,180 patients + 402 caregivers[a]	Survey	Patient and Caregiver QoL	None (qualitative responses)
Heatwole 2016[9]	US	70	Observational	Patient QoL	MDHI, SF-36, INQoL, ESS
Heatwole 2018[10]	US	52	Observational	Patient QoL	MDHI, SF-36 (v2), INQoL
Heatwole 2021[11]	US	40	Clinical study	Patient QoL	MDHI
Holmøy 2019[12]	Norway	22	Observational	Patient QoL and Mental Health	EQ-5D-3L, NPCS
Kurauchi 2019[13]	Japan	43	Observational	Patient and Caregiver QoL, Patient Mental Health	Patients: SF-36v2, CES-D, ESS Caregivers: ZBI
Laberge 2013[14]	Canada	200	Observational	Patient QoL and Mental Health	DSS, SCL-90-R, SF-36

Author, Year	Country	Sample Size	Study Type	Type of Humanistic Burden Reported	Key Instruments Used
Landfeldt 2019[15]	UK	60	Observational	Patient QoL and Mental Health	INQoL, MMSE
Miller 2021[16]	US & UK	39	Observational	Patient Mental Health	BDI, AES, SCOPA-Sleep
MDF 2017[17]	US	26 patients + 26 caregivers	Survey	Patient and Caregiver QoL	None (qualitative responses)
Peric 2013[18]	Serbia	120	Observational	Patient QoL and Mental Health	MSPSS, MIRS, AIS, Ham-D, KFSS, DSS, SF-36
Okkersen 2018[19] van As 2021[20]	France, Germany, Netherlands, UK	255	Clinical study	Patient QoL	INQoL, BDI, DM1-Activ-c
Van Heugten 2018[21]	Netherlands	66	Survey	Patient Mental Health	AES, HADS

AES = Apathy Evaluation Scale; AIS = acceptance of illness scale; BDI = Beck Depression Inventory; CAT = Clinical Assessment for Attention; CES-D = Center for Epidemiologic Studies Depression Scale; DM = myotonic dystrophy; DM1 = myotonic dystrophy type 1; DM1-Activ-c = Myotonic Dystrophy type 1 Activity and participation scale; DSS = Daytime Sleepiness Scale; ENB-2 = Esame Neuropsicologico Breve 2; EQ-5D = EuroQol–5 Dimensions; EQ-5D-3L = EuroQol–5 Dimensions–3 Levels; ESS = Epworth Sleepiness Scale; FAB = Frontal Assessment Battery; GIQLI = Gastrointestinal Quality of Life Index; HADS = Hospital Anxiety and Depression Scale; Ham-D = Hamilton rating scale for depression; INQoL = Individualized Neuromuscular Quality of Life; KFSS = Krupp's Fatigue Severity Scale; LARS = Lille Apathy Rating Scale; MDHI = Myotonic Dystrophy Health Index; MDQoL = Muscular Dystrophy Quality of Life Scale; MFI = Multidimensional Fatigue Inventory; MINI = Mini International Neuropsychiatric Interview; MIRS = Muscular Impairment Rating Scale; MMSE = Mini Mental State Examination; MSPSS = Multidimensional Scale of Perceived Social Support; NPCS = Needs and Provisions Complexity Scale; PHQ-9 = Patient Health Questionnaire–9; SCL-90-R = Symptom Checklist–90–Revised; SCOPA-Sleep = Scales for Outcomes in Parkinson's Disease–Sleep; SF-36 = Short Form–36; SRS = Social Responsiveness Scale; TMT = Trail-Making Test; ZBI = Zarit Caregiver Burden Interview.

[a] Included all types of DM, 39% confirmed DM1

Most Common Instruments

Table 2: Most Common QoL Instruments (Used in ≥2 Studies)

Instrument	Instrument Details
Most Common QoL Instruments	
SF-36	• Generic measure of health-related QoL, with 36 items across 8 domains (physical function, role limitations due to physical function, bodily pain, general health, vitality, social function, role limitations due to emotional function, and mental health), as well as two summary scores for physical components (PCS) and mental components (MCS)[6,15] • Scores are normalized to 0-100, with 0 representing poor QoL and 100 representing optimal QoL[14]
INQoL	• 10-item assessment of muscle disease-related QoL, including the burden of motor impairment and psychosocial limitations[1] • Section and total scores are calculated as percentages, with higher values representing worse burden[1]
EQ-5D	• Generic measure of health-related QoL with 5 domains (mobility, self-care, usual activities, pain, and anxiety/depression) as well as a VAS to assess general health state[12] • Domain scores are transformed into an index value from 0 to 1 and VAS scores range from 0 to 100, with 0 representing the worst possible QoL and 100 representing the best possible QoL[3,12]
Most Common Physical Health Instruments	
MDHI	• DM1-specific instrument including 17 subscales relating to major symptomatic themes in this population, including (but not limited to) mobility, upper extremity function, fatigue, and pain[9] • Total scores range from 0 to 100, with higher values representing worse disease burden[9]
Most Common Mental Health Instruments	
BDI	• Likert scale-based questionnaire measuring symptoms of depression[16] • Scores range from 0 to 63, with higher values representing greater severity[16]
AES	• 18-item assessment of self-reported apathy[16] • Scores range from 18 to 63, with higher values representing greater apathy[16]
CES-D	• Questionnaire focusing on symptoms of depression, with 20 items rated on a 5-point scale[13] • Scores range from 0 to 60, with higher values representing greater depression and a total score ≥16 implying the possibility of depression[5,13]
SCL-90-R	• 90-item scale measuring primary psychological symptoms across 9 dimensions, with 3 global indices (Global Severity Index, Positive Symptom Distress Index, Positive Symptom Total)[14] • Dimension and index scores are transformed into T-scores, which show how much a patient diverges from the normal range for the general population[4,14]

AES = Apathy Evaluation Scale; BDI = Beck Depression Inventory; CES-D = Center for Epidemiologic Studies Depression Scale; EQ-5D = EuroQol–5 Dimensions; INQoL = Individualized Neuromuscular Quality of Life; MCS = Mental Component Summary; MDHI = Myotonic Dystrophy Health Index; PCS = Physical Component Score; QoL = quality of life; SCL-90-R = Symptom Checklist–90–Revised; SF-36 = Short Form–36; VAS = visual analog scale

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