Table 3. Characteristics of included studies – Diagnostiek respiratoire complicaties bij myotone dystrofie

Study	Participants	Data collection	Duration of follow-up	Outcome measures	Results	Risk of bias[1]
Boussaid 2018, Neuromuscular disorders Prospective observational cohort study Country: France	N: 283 Age (mean, SD): I: 43.7 ± 12.5 Sex (% male) I: 57% All participants had Myotonic Dystrophy and were followed-up at the home ventilation unit.	Data were collected prospectively during the first respiratory function evaluation then during the usual annual follow-up evaluations,	for 5 years or until the patient was lost to follow-up.	CTG repeat number, and lung function test (LFT) results (vital capacity [VC] in the upright and supine positions, maximal inspiratory pressure [MIP], maximal expiratory pressure [MEP], and peak cough flow)	Higher CTG repeat number was associated with peak cough flow impairment ($p = 0.007$) and with lower values for maximal inspiratory pressure ($p < 0.0001$) and upright vital capacity. A vital capacity decline over time was associated with older age at first evaluation ($p < 0.0001$), higher CTG repeat number ($p < 0.0001$), and higher baseline body mass index ($p = 0.0004$).	High (observational cohort study)[1]
Hartog, 2021 Retrospective cohort study Country: Canada	N: 110 Age: 42.92±12.34 Sex: 47% M Records of patients diagnosed with DM1 in Neurology Department	Respiratory symptoms and pulmonary function were assessed during routine clinical evaluations by a single respiratory therapist in the neuromuscular clinic.	4.37±3.01 years	Factors influencing respiratory dysfunction at baseline were investigated with Pearson correlations.	We examined various parameters of respiratory functional status, and found FVC (% predicted) correlated best with other measures of disease severity. Annual change in FVC was −1.42 (std error = 0.381). Greater CTG repeat size, higher MIRS rating, and longer disease duration were all correlated with lower baseline FVC but not with annual rate of change.	High (retrospective cohort study)

	of Ohio State university medical center					
Lizio (2023) Retrospective cohort study Country: Italy	N: 151 Age: 44 [35.0 – 53.0] Sex: 44% male	Demographic, clinical, and anthropometric characteristics were collected, as well as arterial blood gas (ABG) analysis, spirometry, respiratory muscle strength, cough efficacy, and nocturnal oximetry as respiratory assessments			76 had an indication for NIV initiation (50.33%). ABG, spirometry, and nocturnal oximetry prediction models resulted in an excellent discriminatory ability in distinguishing patients who needed NIV from those who did not (AUC of 0.818, 0.808, and 0.935, respectively). An easy-to-use calculator was developed to automatically determine a score of NIV necessity based on the prediction equations generated from each aforementioned prediction model	High (retrospective cohort study)
Rossi (2019) Retrospective multicenter cross-sectional cohort study Country: Italy	N: 268 Age (mean, SD): 46.2 (12.9) Sex (% male) DM1: 56% All participants had received the molecular diagnosis of DM1; each	Available clinical and diagnostic data were used.	Mean follow-up was 119.74 ± 151.14 months (median 97.5).	% Explained variance	CTG expansion size in leukocytes, clinical muscle severity, ost functional parameters of respiratory muscle involvement, presence of cardiac conduction disturbances, pacemaker (PMK), exertion dyspnea, obstructive sleep apnea, and indication and compliance to NIV were all significantly associated with restrictive syndrome at the univariate analysis. In the multivariate model only MIRS and (CTG)n resulted independent predictors.	High (observational cohort study)[1;] [no] internal or external validation

	patient included had performed at his/her Referring Center at least one clinical evaluation together with one pulmonary function test (PFT) by a pneumologist during the last 18months				By multivariate analysis, only MIRS and (CTG)n resulted	
Thil, 2017 Retrospective cohort study	N: 80 Age: 37.7 ± 12.5 Sex 41% male	Intervention: pulmonary function tests (at least 2 evaluations in 5 years)	9.02 ± 3.4 year follow-up	Pulmonary function parameters [forced vital capacity (FVC), forced expiratory volume in 1 second (FEV1), TLC, maxi mal expiratory flow at 25%–75% of FVC (MEF25 75%) and residual volume (RV)] between DM1 patients	22 (27.5%) had restricted respiration at inclusion, 32 (40%) were hypoxemic, and 19 (24%) were hypercapnic. The figures at final evaluation were 52 (65%), 50 (63%), and 19 (24%), respectively. Annual declines in FVC (P < 0.01; b 5–30.27), FEV1 (P < 0.04; b 5–1.06), and TLC (P < 0.03; b 5–15.26) were positively associated with BMI.	High (retrospective cohort study)
Vivekanada (2019) Retrospective cohort Country: UK	N: 126 Age: 45 ± 1.3 Sex: 39% male	Data were collected on their age at enrolment, sex, semiquantitative analysis of CTG repeat expansion with Southern blot analysis, age of first symptom (muscle weakness or myotonia), forced vital capacity (FVC) measurement at first visit, and Muscular Impairment Rating Scale (MIRS) score recorded by a neurologist.	Cross-sectional study	The MIRS is an ordinal 5-point rating scale established in accordance with the clinically recognized distal to proximal progression of muscular involvement in DM113: no muscular impairment (1), minimal signs of weakness (2), distal weakness (3), mild to moderate proximal weakness (4), severe proximal	MIRS and FVC were negatively correlated to nocturnal ventilatory requirements (NPAP). 22 out of 54 patients prescribed NPAP did not tolerate this due to various reasons:	High: observational cohort study

Gutschmidt (2023) Cross-sectional cohort study Country: Germany	N: 172 DM1: 74 | DM2: 72 | healthy control C 26 Age (mean, SD): DM1: 443.84(12.01): C: 38.16 (13.85) Sex (% male) DM1: 57% | C: 42% All participants had confirmed Myotonic Dystrophy type I or type 2 or were healthy volunteers; we focused on type 1	Intervention: Patients filled in the Total SUM score Respicheck (score 0–27) as well as the symptom-based (categorial) score Respicheck CAT (score 0–9) were used. Control: Pulmonary function assessments consisted of spirometry (forced vital capacity, FVC) and manometry (maximal inspiratory pressure, MIP; maximal expiratory pressure, MEP) in a sitting position, adjusted for sex and age, the FVC, MIP and MEP are presented in the predicted% of normal.	Cross-sectional, respicheck before pulmonary function assessments	Respicheck score:, it consists of nine symptom categories with three questions each, recording clinical symptoms that indicate respiratory involvement, resulting in 27 questions with dichotomous answering options (orthopnea, dyspnea when performing activities of daily living, poor sleep, morning headache, apnea, decreased cognitive performance, fatigue, excessive daytime sleepiness, treated chest infections). And spirometry (FVC) and manometry (MIP; MEP); presented in the predicted% of normal.	For DM1, the Respicheck SUM score correlated significantly with the results of the lung function assessment in FVC %pred (p = 0.013; r = −0.287), as well as the Respicheck CAT score with FVC %pred (p = 0.006; r = −0.314) and MIP %pred (p = 0.046; r = −0.234). DM2 Respicheck SUM score significantly correlated with MEP %pred (p = 0.032; r = −0.254).	High (observational cohort study)[1]
De Mattia 2020 Country: Italy	N: 58 DM1: 58 Age (mean, SD): DM1: 42[37.0 – 52.0] Sex (% male)	Intervention: The Respicheck was assessed for reliability including internal consistency and reproducibility. The internal consistency of the questionnaire was assessed using Cronbach's alpha. Forty-four patients were asked to complete the Respicheck at baseline and after 3 weeks.	Cross-sectional, respicheck and pulmonary function assessments	Internal consistency and correlation with respiratory impairment	Considering the analysis regarding the internal consistency of the Respicheck questionnaire, a Cronbach's alpha of 0.7 was reported. Respicheck total scores significantly correlated with the severity of respiratory impairment based on the laboratory and clinical classification (overall p =	High (observational cohort study)[1]

	DM1: 47% All participants had genetically confirmed Myotonic Dystrophy type I	Control: Forced spirometry (FVC sitting and supine and the difference between these two measures) and respiratory muscle pressures (MIP, MEP) assessments were performed according to the American Thoracic Society (ATS) standards			0.0105). The higher scores were obtained from patients with the worst respiratory health status And QoL perception.	