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Respiratory function and evaluation in individuals with facioscapulohumeral muscular dystrophy in the Muscular Dystrophy Surveillance, Tracking and Research Network[★]

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Abstract

Using data from the US population-based, multisite Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet), we describe respiratory testing and insufficiency among people with facioscapulohumeral muscular dystrophy (FSHD) diagnosed during 2008-2016. We calculated frequencies and proportions for selected outpatient respiratory assessments (pulmonary function tests [PFTs], forced vital capacity (FVC), inspiratory/expiratory pressure, and polysomnograms) and abnormal test results. We examined frequencies by disease characteristics (FSHD type, ages of onset, non-ambulatory status, scoliosis, lordosis), obesity, and number

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Ethical publication statement

We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

CRedit authorship contribution statement

Katherine D Mathews: Writing – original draft, Methodology, Conceptualization. **Jonathan Suhl:** Writing – original draft, Formal analysis, Conceptualization. **Kristin M Conway:** Writing – review & editing, Methodology, Funding acquisition, Conceptualization. **Amy Moore:** Writing – review & editing, Formal analysis, Conceptualization. **Joyce T. Alese:** . **Russell J Butterfield:** Writing – review & editing, Methodology, Funding acquisition. **Paul A Romitti:** Writing – review & editing, Methodology, Funding acquisition, Conceptualization.

Supplementary materials

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of health encounters. Of 170 people with FSHD, 20.0% underwent PFTs during 2008-2016. Polysomnograms were infrequent (14.1%). FVC <80% predicted was recorded for 64.7% of people tested; additional respiratory outcomes were rare (<5%). Frequency of evaluations and respiratory insufficiency were higher among those with known risk factors and longer follow-up. We observed low proportions of respiratory testing among all confirmed cases of FSHD, but relatively high proportions of mild respiratory insufficiency among those tested. The higher proportions of testing among people with conditions that increase risk of respiratory complications suggest targeted monitoring. Broad implementation of the FSHD guidelines recommending all individuals receive baseline respiratory evaluation at diagnosis could identify respiratory insufficiency as a complication of FSHD.

Keywords

Muscular dystrophy; Facioscapulohumeral respiratory insufficiency; Guidelines; Surveillance

1. Introduction

Respiratory insufficiency (restrictive lung disease, impaired cough) is a potential complication of FSHD [1]. Evaluation of respiratory function among those with FSHD typically relies on measures of forced vital capacity (FVC), maximal inspiratory pressure (MIP), maximal expiratory pressure (MEP), and polysomnograms/sleep studies. Most studies reporting on these measures have been small clinical samples conducted internationally [2–7]. Findings have been mixed with some studies reporting impaired respiratory function, [6–9] but others reporting normal respiratory function among most study participants [2,4,5,10]. Decreased respiratory function has been consistently noted among people with more severe FSHD symptoms or longer disease duration [2,4]. Only two studies were identified that examined early onset (<18 years of age) FSHD, with both reporting impaired respiratory function in this patient population [11,12]. A recent single-site US study reported nearly 50% of people tested showed reduced FVC and recommended continued study of respiratory complications in FSHD using population-based samples to improve generalizability [13].

There are few studies on respiratory insufficiency in individuals with FSHD in the US, and the sampling for several of the studies included people with risk factors known to increase risk of respiratory insufficiency [3–5,7,10,13,14]. Further characterization of respiratory dysfunction in this population early in disease confirmation will give additional insights into the monitoring and management of respiratory function in these individuals, in particular baseline assessments of respiratory function [1]. As such, we used data from the US population-based Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet) to describe patterns and results of respiratory monitoring of people with FSHD.

2. Methods

2.1. Surveillance data collection

MD STAR^{net} surveillance data were collected at each site under Institutional Review Board approval/exemption or public health authority. MD STAR^{net} methods have been described previously [15]. Briefly, people with an eligible muscular dystrophy diagnosis (Becker, congenital, distal, Duchenne, Emery-Dreifuss, FSHD, limb-girdle, myotonic) diagnosed and treated during 2008-2016 who resided in Colorado (CO), Iowa, (IA), western New York (21 counties, [NY]), North Carolina (33 counties in the Piedmont region [NC]), South Carolina (SC), or Utah (UT)/Nevada (NV) were identified.

2.2. Diagnosis of FSHD

Using a standardized abstraction tool, trained abstractors collected diagnostic and clinical data from medical records for all available health encounters during 2008-2016. Clinical signs and symptoms of progressive muscle weakness involving the face, scapular stabilizers, upper arm, lower leg (peroneal muscles), and hip girdle, or language commonly used to describe these symptoms, were collected from the medical record and included: dysarthria, eye closure weakness, facial weakness, family history, foot drop/foot weakness, lordosis, scapular winging, shoulder weakness, trouble climbing up and down stairs, trouble raising arm, trouble sitting up from lying, trouble smiling, trouble walking/running/jumping, trouble whistling/drinking from straw, truncal/abdominal weakness, and hearing loss or retinopathy (children). The dates (month/year) or age (years/months) of onset for each symptom was recorded as reported in the medical record. Confirmatory DNA test results (4q35 D4Z4 contraction size, 4qA/4qB allele size) and details about family history, including inheritance pattern, were also collected from the medical record, where available.

A clinical review committee reviewed clinical signs and symptoms and diagnostic testing results to assign standardized case classifications of definite, probable, possible, or asymptomatic and FSHD subtype (FSHD1, FSHD2, FSHD NOS) (Supplemental Box 1). Two clinicians (K.D.M, R.J.B.) also assigned early onset FSHD using published criteria for onset ages of facial weakness (<5 years old) and shoulder or proximal weakness (<10 years old) [16].

2.3. Sociodemographic and clinical variables

Sociodemographic variables examined were race/ethnicity (non-Hispanic White/non-Hispanic Black/Hispanic/Other or Unknown), sex (male/female), and MD STAR^{net} site (CO, IA, wNY, NC, SC, UT). The following clinical parameters were determined at diagnosis or at any time during followup: known FSHD family history (yes/no), early FSHD onset (child/adult/undetermined), mobility status (ambulatory/non-ambulatory [full-time wheelchair/documentary ceased ambulation]) and presence of scoliosis (cobb angle $\geq 20^\circ$ /no spinal x-ray or cobb angle $< 20^\circ$), lordosis (present/not documented), and obesity (BMI $> 30 \text{ kg/m}^2$ [≥ 20 years] or $\geq 95^{\text{th}}$ percentile CDC growth chart[17], 2-19 years).

2.4. Respiratory testing

Respiratory testing data collected at clinical encounters during the surveillance period (2008-2016) were extracted from medical records. Data from abstracted pulmonary function tests (PFTs) included FVCs and MIP/MEP expiratory pressures. For each PFT, the date of the test, the type of test (FVC, MIP, MEP) and the test results were collected. For FVC, FVC percent predicted or FVC absolute (in liters) were abstracted, and cm H₂O were collected for MIP and MEP. Impaired respiratory function was defined as any FVC <80% or <50% of predicted; an absolute FVC <1L; and MIP <60cmH₂O or MEP <80cmH₂O noted in any PFT during follow-up. Physician notes for PFTs were also abstracted and reviewed. Nocturnal hypoventilation was defined as a noted diagnosis of nocturnal hypoventilation or identification through clinical review of test results of polysomnogram, sleep study, or overnight oximetry, or symptom description (full time or nighttime NIPPV or oxygen use). To evaluate opportunity for respiratory testing, we also classified people as having <4 or ≥4 encounters with a neuromuscular/neurology specialist during 2008-2016 [6].

2.5. Statistical analysis

We calculated descriptive statistics for continuous and categorical variables. Proportions and 95% Wilson score confidence intervals were calculated for the total sample and a subsample of persons with documented respiratory testing as the denominator. Additional proportions include stratification by any documented risk factor for respiratory compromise (obesity, mobility status, scoliosis, lordosis, or early FSHD onset), each individual risk factor, and number of encounters (<4 vs ≥4). Small numbers precluded statistical testing to evaluate comparisons. Analyses were conducted using SAS v 9.4 [18].

2.6. Inclusions and exclusions

For this study, people with genetically confirmed FSHD (definite) or those with clinical symptoms of FSHD and family history of FSHD consistent with a dominant inheritance (probable) were eligible for analysis (people with possible FSHD were not pooled for analyses). Additionally, data from NV were excluded from analysis due to incomplete ascertainment and follow-up.

3. Results

Overall, 173 people with FSHD were included in the MD STAR_{net}, of which 170 people met the inclusion criteria. Most were confirmed by genetic testing, diagnosed with FSHD1, and classified as adult-onset (Table 1). The sample was largely non-Hispanic white and evenly distributed between males and females. Most had known family history of FSHD, remained ambulant, and had no documentation of scoliosis or lordosis during follow-up. Nearly one-third could be classified as ever obese. Average ages of first and last health encounters captured by MD STAR_{net} were 39 (*SD*=21.0) and 42 (*SD*=21.3) years, respectively; median follow-up time was 4 years (minimum = 1 year, maximum = 9 years; IQR = 2-6 years). Demographic characteristics tended to be similar between those with early- and adult-onset FSHD; proportions for presence of risk factors for respiratory insufficiency tended to be higher for early-onset (Table 2).

One in five people had FVC testing; other respiratory tests were less frequent (Table 3). The first FVC testing occurred within an average of 1.9 ($SD = 1.8$) years of their first health encounter. The median FVC at initial documented testing was 77% of predicted; median MIP and MEP were 58.0 cmH₂O and -50.0 cmH₂O, respectively. At least mildly impaired respiratory function (<80% FVC) was noted for a small percentage of people overall but for the majority of those tested. No individual had a recorded FVC <1.0L.

Higher proportions of respiratory testing were observed among those with at least one risk factor for respiratory compromise (Table 4) and for each factor individually (Table 5) than people without. Conversely, the proportion of people with any abnormal respiratory function test was generally as high or higher among people without any risk factors compared to people with at least one risk factor (Table 4). Proportions of people who had respiratory testing were higher among those with four or more neurology/neuromuscular visits (FVC=32.4%, MIP/MEP=16.9%, polysomnogram=19.7%) compared to people with fewer than four visits (FVC=11.1%, MIP/MEP=4.0%, polysomnogram=10.1%) (data not shown).

4. Discussion

Using a US population-based, multisite sample, we found 20% of people with FSHD had documented respiratory testing during 2008-2016. Although we had a small number of people with known risk factors for respiratory insufficiency and found more frequent evaluations among these people, the proportion of people tested was lower than would be expected given longstanding recognition of the importance of pulmonary monitoring for at-risk patients. Our study supports the current recommendations of baseline respiratory testing in FSHD given our finding of impaired respiratory function among people without known risk factors for respiratory insufficiency.

Previous studies of respiratory function of people with FSHD have reported variable rates of abnormal test results ranging from less than 10% to near 50% [2,4–13]. The variability of reported findings is likely due in part to disease stage of the underlying population, with more severely affected patient populations demonstrating greater respiratory insufficiency [2–6,9–12]. Unlike most published series, our sample is restricted to people with confirmation of an FSHD diagnosis during the surveillance period, possibly skewing to a younger population and milder stage of disease. Because our sample is retrospective and a minority of patients underwent testing, we cannot estimate the frequency of respiratory compromise. However, we note that among those tested, we observed at least mild respiratory insufficiency in more than one-half.

Interestingly, we observed similar frequencies of at least mildly abnormal FVCs among tested people with and without risk factors. It is possible that other clinical factors prompted testing in those without the risk factors we considered here. Alternatively, mild respiratory compromise might be common and unaffected by the studied risk factors. A 5-year follow up study of respiratory function in 92 people with FSHD showed a mean baseline FVC of 79% predicted. Rapid decline in FVC was seen in a minority and associated with stage of disease (weakness) and spinal deformity within 5 years [6]. Although only a small

proportion of our sample had MIP/MEP testing, approximately one-half were abnormal. This suggests that impaired airway clearance and increased aspiration risk might be more common than recognized in FSHD and merits screening questions, clinical assessment, and selected formal testing. We note that peak cough flow, another common measure of airway clearance, was too infrequent (n=8) to analyze separately and MD STAR_{net} did not collect data on insufflator/exsufflator use or symptoms (e.g., frequent or prolonged respiratory infections) that may indicate impaired airway clearance in people with FSHD.

FSHD is usually a relatively slowly progressing disorder. Some with the disease might have limited routine healthcare, particularly as it relates to FSHD. We observed that those who had more frequent neurology/neuromuscular provider visits during the surveillance period were more likely to have respiratory function testing recorded than those with less frequent visits. This difference suggests that those who have more specialized healthcare visits may be more likely to have care that allows monitoring of respiratory symptoms or risk factors that may prompt respiratory testing. Alternatively, those with more advanced disease or those experiencing symptoms related to respiratory dysfunction might be more likely to seek care and thus be tested. Finally, it could be that those with fewer documented visits, or less centralized care, are receiving care at clinic sources not accessible to the MD STAR_{net}. Available MD STAR_{net} data do not allow us to distinguish these possibilities.

Study limitations include reliance on available medical records and small numbers limiting statistical comparisons. We did not collect indications (e.g., shortness of breath, symptoms of nocturnal hypoventilation [morning headaches, feeling sleepy in the daytime, not feeling refreshed in the morning, dizziness]) for respiratory testing among those tested. It is possible that respiratory symptoms reported in clinic, but not included as part of our risk profile, prompted some evaluations. This may have biased initial test results towards evidence of respiratory insufficiency. In addition, routine FVC testing can be physically challenging and inaccurate due to disease-related facial weakness; thus, some people might have attempted testing, but valid results could not be determined. MD STAR_{net} does not collect severity of signs and symptoms (e.g, facial weakness) of FSHD. Thus, we were unable to identify these situations, and such people would have been considered untested if the attempt was not recorded in the medical record and abstracted [3,6]. In addition, MD STAR_{net} does not collect data on the method used for pulmonary testing (mask vs mouthpiece, for example) that might affect the accuracy of the test result. Testing quality was not documented for all respiratory testing.

5. Conclusion

Our study found that few people with newly diagnosed FSHD have routine respiratory function screening but a substantial proportion of those tested showed evidence of respiratory insufficiency. Additionally, although people with risk factors for respiratory complications were more likely to be tested, respiratory insufficiency was identified among people without these risk factors at similar or even higher proportions than found among those at risk. Combined with a recent MD STAR_{net} cross-sectional study that found respiratory complications were a contributing factor to death in over 50% of deceased individuals with FSHD, [19] these findings suggest respiratory insufficiency may

be a frequent complication of FSHD regardless of perceived risk level. In addition to recommended baseline testing, our findings suggest regular respiratory monitoring of people with FSHD should be considered, even in the absence of classic signs and symptoms of respiratory insufficiency [1,4]. Future studies are needed to establish the interval at which monitoring should occur.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

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Declaration of competing interest

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Data availability

Due to privacy concerns, data from the MD STAR_{net} are not publicly available. Researchers interested in MD STAR_{net} data can contact MDSTARnet@cdc.gov. Data used for this analysis are maintained at The University of Iowa MD STAR_{net} site.

Abbreviations

MD STAR_{net}	Muscular Dystrophy Surveillance Tracking and Research Network
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References

- [1]. Tawil R, Kissel JT, Heatwole C, Pandya S, Gronseth G, Benatar M, et al. Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy: report of the guideline development, dissemination, and implementation subcommittee of the American Academy of Neurology and the practice issues review panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. *Neurology*, 2015;85(4):357–64. 10.1212/WNL.0000000000001783. [PubMed: 26215877]
- [2]. D'Angelo MG, Romei M, Lo Mauro A, Marchi E, Gandossini S, Bonato S, et al. Respiratory pattern in an adult population of dystrophic patients. *J Neurol Sci* 2011;306(1-2):54–61. 10.1016/j.jns.2011.03.045. [PubMed: 21529845]

- [3]. Henke C, Spiesshoefer J, Kabitz HJ, Herkenrath S, Randerath W, Brix T, et al. Respiratory muscle weakness in facioscapulohumeral muscular dystrophy. *Muscle Nerve* 2019;60(6):679–86. 10.1002/mus.26717. [PubMed: 31566774]
- [4]. Moreira S, Wood L, Smith D, Marini-Bettolo C, Guglieri M, McMacken G, et al. Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. *J Neurol* 2017;264(6):1271–80. 10.1007/s00415-017-8525-9. [PubMed: 28550484]
- [5]. Stubgen JP, Schultz C. Lung and respiratory muscle function in facioscapulohumeral muscular dystrophy. *Muscle Nerve* 2009;39(6):729–34. 10.1002/mus.21261. [PubMed: 19301367]
- [6]. Teeselink S, Vincenten SCC, Voermans NC, Groothuis JT, Doorduyn J, Wijkstra PJ, et al. Long-term follow-up of respiratory function in facioscapulohumeral muscular dystrophy. *J Neurol* 2022;269(7):3682–9. 10.1007/s00415-022-10990-7. [PubMed: 35147730]
- [7]. Wohlgenuth M, van der Kooi EL, van Kesteren RG, van der Maarel SM, Padberg GW. Ventilatory support in facioscapulohumeral muscular dystrophy. *Neurology*, 2004;63(1):176–8. 10.1212/01.wnl.0000133126.86377.e8. [PubMed: 15249635]
- [8]. Wohlgenuth M, Horlings CGC, van der Kooi EL, Gilhuis HJ, Hendriks JCM, van der Maarel SM, et al. Respiratory function in facioscapulohumeral muscular dystrophy 1. *Neuromuscul Disord* 2017;27(6):526–30. 10.1016/j.nmd.2017.03.008. [PubMed: 28416348]
- [9]. Santos DB, Boussaid G, Stojkovic T, Orlikowski D, Letilly N, Behin A, et al. Respiratory muscle dysfunction in facioscapulohumeral muscular dystrophy. *Neuromuscul Disord* 2015;25(8):632–9. 10.1016/j.nmd.2015.04.011. [PubMed: 26023000]
- [10]. Scully MA, Eichinger KJ, Donlin-Smith CM, Tawil R, Statland JM. Restrictive lung involvement in facioscapulohumeral muscular dystrophy. *Muscle Nerve* 2014;50 (5):739–43. 10.1002/mus.24218. [PubMed: 24639337]
- [11]. Trucco F, Pedemonte M, Fiorillo C, Tacchetti P, Brisca G, Bruno C, et al. Respiratory pattern in a FSHD pediatric population. *Respir Med* 2016;119:78–80. 10.1016/j.rmed.2016.08.014. [PubMed: 27692152]
- [12]. Goselink RJM, Mul K, van Kernebeek CR, Lemmers R, van der Maarel SM, Schreuder THA, et al. Early onset as a marker for disease severity in facioscapulohumeral muscular dystrophy. *Neurology* 2019;92(4):e378–85. 10.1212/WNL.0000000000006819. p. [PubMed: 30568007]
- [13]. Kelly CR, Saw JL, Thapa P, Mandrekar J, Naddaf E. Systemic manifestations and symptom burden of facioscapulohumeral muscular dystrophy in a referral cohort. *Muscle Nerve* 2022;65(4):415–21. 10.1002/mus.27493. [PubMed: 35020192]
- [14]. Kilmer DD, Abresch RT, McCrory MA, Carter GT, Fowler WM, Johnson ER Jr, et al. Profiles of neuromuscular diseases. Facioscapulohumeral muscular dystrophy. *Am J Phys Med Rehabil* 1995;74(5):S131–9. 10.1097/00002060-199509001-00007. Suppl. [PubMed: 7576420]
- [15]. Kang PB, Jorand-Fletcher M, Zhang W, McDermott SW, Berry R, Chambers C, et al. Genetic patterns of selected muscular dystrophies in the muscular dystrophy surveillance, tracking, and research network. *Neurol Genet* 2023;9(6):e200113. 10.1212/NXG.000000000000200113. [PubMed: 38045992]
- [16]. Brouwer OF, Padberg GW, Wijmenga C, Frants RR. Facioscapulohumeral muscular dystrophy in early childhood. *Arch Neurol*, 1994;51(4):387–94. 10.1001/archneur.1994.00540160085011. [PubMed: 8155016]
- [17]. Kuczmarski RJ, Ogden CL, Guo SS, Grummer-Strawn LM, Flegal KM, Mei Z, et al. 2000 CDC growth charts for the United States: methods and development. *Vital Health Stat* 2002;11(246):1–190.
- [18]. SAS Institute Inc., SAS 9.4 Cary, NC: SAS Institute Inc.; 2013.
- [19]. Wallace B, Smith KT, Thomas S, Conway KM, Westfield C, Andrews JG, et al. Characterization of individuals with selected muscular dystrophies from the expanded pilot of the Muscular Dystrophy Surveillance, Tracking and Research Network (MD STARnet) in the United States. *Birth Defects Res* 2020. 10.1002/bdr2.1764.

Table 1Demographic and clinical characteristics of 170 people with FSHD ascertained by MD STAR_{net}, 2008-2016.

Characteristics	N (%)
Case Classification	
Genetically confirmed	127 (74.7)
Dominant family history, without genetic testing	43 (25.3)
FSHD Clinical Subtype	
FSHD1	126 (74.1)
FSHD2	2 (1.2)
FSHD NOS	42 (24.7)
Age at FSHD Onset	
Early	14 (8.2)
Adult	123 (72.4)
Undetermined	33 (19.4)
Race/Ethnicity	
Non-Hispanic White	138 (81.2)
Other	32 (18.8)
Sex	
Male	96 (56.5)
Female	74 (43.5)
Known Family History	
No	51 (30.0)
Yes	119 (70.0)
Mobility Status	
Ambulatory	163 (95.9)
Non-ambulatory	7 (4.1)
Scoliosis	
No	160 (94.1)
Yes	9 (5.3)
Unknown	1 (0.6)
Lordosis	
No	134 (78.8)
Yes	36 (21.2)
Obesity	
No	112 (65.9)
Yes	48 (28.2)
Unknown	10 (5.9)

MD STAR_{net}: Muscular Dystrophy Surveillance, Tracking and Research Network.

Table 2

Demographic and clinical characteristics of 137 people by early- or adult-onset FSHD ascertained by MD STAR^{net}, 2008-2016.

Characteristics	Early Onset (N=14)	Adult Onset (N=123)
	N (%)	N (%)
FSHD Clinical Subtype		
FSHD1	12 (85.7)	93 (75.6)
FSHD2	2 (14.3)	28 (22.8)
FSHD NOS	0 (0.0)	2 (1.6)
Sex		
Male	7 (50.0)	73 (59.4)
Female	7 (50.0)	50 (40.7)
Known Family History		
No	7 (50.0)	36 (29.3)
Yes	7 (50.0)	87 (70.7)
Mobility Status		
Ambulatory	13 (92.9)	118 (95.9)
Non-ambulatory	1 (7.1)	5 (4.1)
Scoliosis		
No	12 (85.7)	118 (95.9)
Yes	2 (14.3)	5 (4.1)
Lordosis		
No	8 (57.1)	103 (83.7)
Yes	6 (42.9)	20 (16.3)
Obesity		
No	11 (78.6)	82 (66.7)
Yes	2 (14.3)	35 (28.4)
Unknown	1 (7.1)	6 (4.9)

MD STAR^{net}: Muscular Dystrophy Surveillance, Tracking and Research Network.

¹Other includes non-Hispanic Black, Hispanic, and other/unknown.

Table 3

Respiratory testing and function for 170 people with FSHD ascertained by MD STAR_{net}, 2008-2016.

		Total Population (n=170)	Tested Population (n=34)
Respiratory Testing and Function	N	% (95% CI)	% (95% CI)
Pulmonary Function Testing			
Any	34	20.0 (14.4, 27.0)	
FVC	34	20.0 (14.4, 27.0)	
MIP/MEP	16	9.4 (5.6, 15.1)	
Polysomnogram	24	14.1 (9.4, 20.5)	
Pulmonary Function			
FVC			
< 80% predicted	22	12.9 (8.5, 19.2)	64.7 (46.5, 79.7) ¹
< 50% predicted	4	2.4 (0.8, 6.3)	11.8 (3.9, 28.4) ¹
MIP (< 60 cm H ₂ O)	8	4.7 (2.2, 9.4)	50.0 (25.5, 74.5) ²
MEP (< - 80 cm H ₂ O)	7	4.1 (1.8, 8.6)	43.8 (20.8, 69.4) ²
Nocturnal hypoventilation	19	11.2 (7.0, 17.1)	NC ³

FVC: forced vital capacity; MD STAR_{net}: Muscular Dystrophy Surveillance, Tracking and Research Network; MEP: maximum expiratory pressure; MIP: maximum inspiratory pressure; NC: not calculated.

¹Denominator=people with FVC testing (n=34).

²Denominator=people with MIP/MEP testing (n=16).

³Nocturnal hypoventilation was determined from polysomnograms completed during follow-up or description of receiving a diagnosis prior to the surveillance period; therefore, the denominator representing people tested is unknown.

Table 4

Respiratory testing and function by any risk factor for 170 people with FSHD ascertained by MD STARnet, 2008–2016.

	No Risk Factor (n=83)				Any Risk Factor (n=87)			
	N	N	% Total (95% CI)	% Tested (95% CI)	N	% Total (95% CI)	% Tested (95% CI)	
Any Pulmonary Function Testing								
Any	34	12	14.5 (8.0, 24.3)		22	25.3 (16.8, 35.9)		
FVC	34	12	14.5 (8.0, 24.3)		22	25.3 (16.8, 35.9)		
MIP/MEP	16	6	7.2 (3.0, 15.6)		10	11.5 (5.9, 20.6)		
Polysomnogram	24	8	9.6 (4.6, 18.6)		16	18.4 (11.2, 28.4)		
Respiratory Outcome								
FVC								
< 80% predicted	22	8	9.6 (4.6, 18.6)	66.7 (35.4, 88.7) ¹	14	16.1 (9.4, 25.9)	63.6 (40.8, 82.0) ¹	
< 50% predicted	4	1	1.2 (0.1, 7.5)	8.3 (0.4, 40.2) ¹	3	3.4 (0.9, 10.5)	13.6 (3.6, 36.0) ¹	
MIP/MEP Testing								
MIP (< 60 cm H ₂ O)	8	4	4.8 (1.6, 12.5)	66.7 (24.1, 94.0) ²	4	4.8 (1.5, 12.0)	40.0 (13.7, 72.6) ²	
MEP (< -80 cm H ₂ O)	7	3	3.6 (0.9, 10.9)	50.0 (13.9, 86.1) ²	4	4.8 (1.5, 12.0)	40.0 (13.7, 72.6) ²	
Nocturnal hypoventilation	19	5	6.0 (2.2, 14.1)	NC ³	14	16.1 (9.4, 25.9)	NC ³	

FVC: forced vital capacity; MD STARnet: Muscular Dystrophy Surveillance, Tracking and Research Network; MEP: maximum expiratory pressure; MIP: maximum inspiratory pressure; NC: not calculated.

¹Denominator=people with FVC testing (n=12 [no risk factor]; n=22 [any risk factor]).

²Denominator=people with MIP/MEP testing (n=6 [no risk factor]; n=10 [any risk factor]).

³Nocturnal hypoventilation was determined from polysomnograms completed during follow-up or description of receiving a diagnosis prior to the surveillance period; therefore, the denominator representing people tested is unknown.

Respiratory testing and results by individual comorbidities for 170 people with FSHD ascertained by MD STAR_{net}, 2008-2016.

Table 5

Respiratory test result	Total population (n=170)	Early onset (n=14)	Non-ambulatory (n=7)	Scoliosis (n=9)	Lordosis (n=36)	Obesity (n=48)
FVC Testing	34 (20%)	5 (35.7%)	3 (42.9%)	4 (44.4%)	11 (30.6%)	12 (25.0%)
< 80% predicted	22 (12.9%)	3 (21.4%)	2 (28.9%)	4 (44.4%)	7 (19.4%)	7 (14.6%)
< 50% predicted	4 (2.4%)	1 (7.1%)	1 (14.3%)	2 (22.2%)	2 (5.6%)	0 (0.0%)
MIP/MEP Testing	16 (9.4%)	3 (21.4%)	2 (28.6%)	2 (22.2%)	5 (13.9%)	5 (10.4%)
MIP (< 60 cm H ₂ O)	8 (4.7%)	0 (0.0%)	1 (14.3%)	2 (22.2%)	3 (8.3%)	2 (4.2%)
MEP (< -80 cm H ₂ O)	7 (4.1%)	1 (7.1%)	2 (28.6%)	1 (11.1%)	2 (5.6%)	2 (4.2%)
Polysomnogram	24 (14.1%)	3 (21.4%)	1 (14.3%)	2 (22.2%)	5 (13.9%)	13 (27.1%)
Nocturnal Hypoventilation	19 (11.2%)	4 (28.6%)	1 (14.3%)	2 (22.2%)	5 (13.9%)	10 (20.8%)

FVC: forced vital capacity; MD STAR_{net}: Muscular Dystrophy Surveillance, Tracking and Research Network; MEP: maximum expiratory pressure; MIP: maximum inspiratory pressure.
 Note. People may be counted in more than one risk factor.