Dyspnea in Dystonia*
A Functional Evaluation

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Background: Dystonia consists of action-triggered sustained focal muscle contractions, worsened by effort, and resulting in voice changes, abnormal posturing, and dyspnea. The cause of dyspnea, previously unexplained, is the basis of this report.

Methods: Since the maximal efforts required to perform pulmonary function testing (PFT) could worsen the muscular contractions in dystonic patients, we used several tests to identify possible causes of dyspnea. These included spirometry with flow volume loops (FVL), tidal volume breathing, maximum voluntary ventilation (MVV), and inspiratory and expiratory muscle pressures (PImax, PEmax), sitting and supine. We used cycle ergometry with arterial blood gas (ABG) values to detect cardiac/pulmonary limitations and respiratory inductive plethysmography (RIP) to assess chest wall/abdominal movements for synchrony. Dynamic videofluoroscopy (VF) assessed and recorded the action-triggered muscle activity of the upper airways and the diaphragm during quiet breathing, speech, swallowing, and maximal respiratory maneuvers similar to the efforts required during PFT.

Results: Twenty-six dystonic patients, 12 women and 14 men, ages 14 to 70 years (mean age, 52.3 years) were evaluated. Their neurologic classification included 22 primary (idiopathic) and 4 secondary (2 postneuroleptic use, 2 posttraumatic). Four patients originally classified as having focal dystonia had dyspnea and were found to have diaphragmatic and/or upper airway dysfunction too. The PFTs showed abnormal FVL and/or tidal volume breathing patterns, with intermittent interruptions of air flow during inspiration or expiration in 20 of 24 patients. The VF was abnormal in 24 of 26 patients: 19 patients had combined upper airway (UA) and diaphragmatic dysfunction (DD); 1 patient had UA dysfunction alone, and 4 patients had DD alone. Except for poor effort and/or dystonic movements, cycle ergometry was normal in 18 of 21 patients. The ABG values and/or pulse oximetry were normal in 19 of 22 patients.

Conclusion: Dyspnea in dystonia appears to be due to excessive and/or dysrhythmic contractions of the upper airways and/or diaphragm, with usually normal gas exchange. These spasmotic and irregular muscular contractions during speech and daily activities are associated with the sensation of excessive effort to overcome the spasms. Excessive spasms can be triggered during PFT and are best detected on FVL patterns coupled with dynamic VF. (Chest 1995; 107:1309-16)

Dystonia is a syndrome dominated by involuntary sustained muscle contractions frequently causing twisting and repetitive movements or abnormal postures. It can involve any voluntary muscle. Because dystonia is rare and its movements and resulting postures are often unusual, it is one of the most frequently misdiagnosed neurologic conditions.1

In this country, the prevalence of all forms of dystonia is estimated to be about 200,000 cases.2 As a clinical syndrome, dystonic patients are classified according to age at onset, symptoms, and etiology. When the presenting signs and symptoms appear before the age of 12 years, it is designated of childhood onset. It is of adolescent onset when signs and symptoms appear between the ages of 13 and 20 years, and of adult onset in older patients.

Dystonia can be primary or without a hereditary pattern, or secondary to a number of conditions (Table I[A]) with focal, multifocal, or generalized manifestations (Table I[B]). Focal dystonia involves one small group or a contiguous group of muscles in one body part. Multifocal dystonia involves two different noncontiguous groups of muscles.

Dyspnea associated with dystonia has been infrequently reported.3 Dyspnea may be due to central nervous system disorders, medications, cardiac or pulmonary disorders, or anxiety. Thus, a comprehensive analysis is necessary to exclude possibilities and determine true cause(s). This report describes the contribution of the upper airways (UA), the lungs, and the diaphragm to dyspnea in patients with dystonia.

Key words: diaphragm; dyspnea; dystonia; pulmonary function testing; upper airways; videofluoroscopy

ABG=arterial blood gas; Dco=diffusion capacity of carbon monoxide; DD=diaphragmatic dysfunction; FVL=flow volume loop; MVV=maximum voluntary ventilation; PEmax=maximal inspiratory muscle pressure; PImax=maximal inspiratory muscle pressure; RIP=respiratory inductive plethysmography; UA=upper airway; VF=videofluoroscopy

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Manuscript received May 12, 1994; revision accepted September 2.
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A. Etiology
1. Primary
   a) Without hereditary pattern
   b) With hereditary pattern (autosomal dominant, X-linked recessive)
2. Secondary
   a) Associated with other hereditary neurologic disorders (eg, Wilson’s and Huntington’s diseases)
   b) Environmental, posttraumatic, postinfectious, postvascular accidents, postdrugs, or toxins
   c) Associated with Parkinson’s disease
   d) Psychogenic
B. Manifestations
1. Focal
   a) Blepharospasm (involuntary blinking)
   b) Orofacial (face, jaw, tongue, affecting swallowing, and/or speech)
   c) Spasmodic torticollis (neck)
   d) Writer’s cramp (action-induced dystonic contraction of hand muscles)
   e) Spasmodic dysphonia (vocal folds, strangled voice)
2. Multifocal
   Cranial, axial, crural
3. Generalized
   Any combination of bilateral lower extremity dystonia with dystonia involving any other body part

METHODS

Twenty-six dystonic patients with dysphonia (strangled quality of speech, with interruptions) and dyspnea were referred for respiratory assessment from the Movement Disorders Center of the Neurological Institute of Columbia Presbyterian Medical Center, New York. Dystonia was primary (idiopathic) in 22 patients and secondary in 4 patients (Table 2). All described an awareness of or an increased effort for breathing. They were assigned a dyspnea score, depending on their perception of difficulty of breathing; as follows: 0, no dyspnea, normal; 1, aware of some dyspnea, but able to fully function; 2, dyspnea with reduced function on exercise; 3, dyspnea with daily activities; and 4, dyspnea at rest and/or painful chest wall/diaphragmatic spasms with daily activities.

After a thorough history and physical examination, all patients underwent pulmonary function testing (PFT). This included spirometry with flow volume loop (FVL), patterns of tidal volume breathing, maximum voluntary ventilation (MVV), maximal inspiratory muscle pressure (Plmax), and maximal expiratory muscle pressure (PEmax) in the sitting and supine positions; lung volumes; diffusion capacity of carbon monoxide (Dco) (Warren E. Collins Inc, Braintree, Mass), arterial blood gases (ABG) (Instrument Laboratory 1306 blood gas analyzer).

Cycle ergometry (a symptom-limited incremental exercise test on a cycle ergometer) was performed to detect any pulmonary or cardiac limitation. Exercise testing was combined with respiratory inductive plethysmography (RIP), (Respiritrace Systems, Ardsley, NY) to detect chest wall/abdominal dysynchrony.

Videofluoroscopy (VF) was performed to evaluate the movements of the pharynx, larynx, and diaphragm during quiet breathing, speech, swallowing, and maximal effort maneuvers (MVV, sniffing and panting), in the erect and supine positions. Each hemidiaphragm was observed in the posteroanterior (PA) and oblique projections to assess its movement and synchrony during these maneuvers. A filter (Thoreus) was used to enhance soft-tissue definition of the upper airways.

Fluoroscopy time was up to 5 min per patient with a total exposure under 12.5 rads. Patient consent was obtained for these tests. After evaluating these patients, similarities among them became apparent. We then requested and obtained approval from the institutional review board to use the data for the purpose of this report. Wilcoxon’s signed rank with Spearman’s correlation were used for statistical analysis with a p value less than 0.02 being considered significant.

RESULTS

There were 12 women and 14 men (Table 2). Their ages ranged from 14 to 70 years (mean, 52.3 ± 13.8). They were grouped into three categories, based on the results of their spirometry and lung volumes: group 1 with normal PFT (n=19), group 2 with obstructive PFT (n=3), and group 3 with restrictive PFT (n=4). Flow volume loops were assessed for interruptions and/or irregularities and noted separately for each group. The etiology and classification of dystonia was determined by the referring neurologist and are listed in Table 2. Four patients were initially classified as having focal dystonia. Patients 4 and 8 had dysphonia and coughing spasms. Patient 17

Figure 1. Tracings from two different patients showing variations in depth and rate of breathing pattern (tidal volume breathing for 35 s).

Figure 2. Highly irregular MVV tracings from two different patients. Both tracings show involuntary breath holding, with occasional grunting (a).
Table 2—Patient Individual Data*

| No./Age, Yrs/Sex | Dystonia Cause | Dystonia Manifest | Dyspnea Score
d | FEV₁/FVC (%Predicted) (S=Smoker) | Resting ABG pH/Pco₂/PO₂/HCO₃ | Plmax/PEmax (% Predicted) | Flow Volume Loop | Exercise Test | Video-fluoroscopy |
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<td>Normal Sat</td>
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<td>NA</td>
<td>Normal</td>
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| Group 2 (n=3)  |                |                   |                 |                               |                           |                  |                 |               |                  |
| 20/52/F         | Primary        | F                 | 2               | 68% (S)                       | 7.44/41/106/26            | 80/80            | Obstr           | Subopt         |                  |
| 21/60/F         | Primary        | G                 | 4               | 64% (S)                       | 7.39/47/72/27             | 106/70           | Obstr and irreg | N (DB)        |                  |
| 22/35/F         | Secondary      | G                 | 4               | 49% (S) (mean: 60.3%) (±10.0%) | 7.43/38/110/26            | 45/55            | Obstr and irreg | Obstr and irreg |                  |

| Group 3 (n=4)  |                |                   |                 |                               |                           |                  |                 |               |                  |
| 23/25/M         | Secondary      | G                 | 1               | 79%                           | 7.40/40/94/25             | 42/40            | Restr and irreg | Subopt+DB      | Abnormal UA and D |
| 24/50/M         | Primary        | G                 | 4               | 77%                           | NA                         | 87/96            | Restr and irreg | NA             | Abnormal UA and D |
| 25/47/M         | Primary        | G                 | 4               | 80% (S)                       | 7.39/47/81/28             | Could not do     | Restr and irreg | NA             | Abnormal UA and D |
| 26/61/F         | Primary        | G                 | 4               | 80% (mean: 79.0%) (±1.4%)     | 7.47/37/98/26             | 39/26            | Irreg           | Normal         | (DB)             |

*F=Focal; MF=multifocal; G=generalized; Sat=saturation; NA=not available; Irreg=irregular; Obstr=obstructive; Restr=restrictive; DB=dystonic breathing; Subopt=suboptimal effort; Card=cardiovascular; Vent=ventilatory; Limit=limitation; D=Diaphragm.

1Dyspnea score, 0-4 (see text).
had occasional gasping and abnormal head movements. All three patients had difficulty swallowing. Patient 20 had blepharospasm, jaw and facial movements, and occasional stridor.

Variation in depth and rate of the breathing pattern was unique to each subject (Fig 1) and was frequently seen in all three groups. Involuntary breath holding with grunting during the MVV maneuver and/or highly irregular MVV (Fig 2) were among the patterns seen. Each of these records is from a different subject.

**Group 1: Normal PFT (n=19)**

Nineteen of the 26 patients (73%) had normal standard spirometry. Two patients did not have FVL available for review. In the remaining 17 patients, FVLs were abnormal in 14 patients (82%) with interruptions during inspiration and/or expiration (Fig 3). Five of these 19 patients (26%) had decreased respiratory muscle pressures with a mean Plmax of 56.2% (±8.3%) of predicted, and a mean PEmax of 50.6% (±7.0%) of predicted (Table 2). For the group as a whole, Plmax was 93.6% (±21.8%) of predicted and PEmax was 87.5% (±25.2%) of predicted.

Fourteen of 16 patients had normal ABG and/or oxygen saturation by pulse oximetry. Two patients had a borderline increased PCO2 (PCO2, 45 mm Hg and 46 mm Hg), one of whom had a PO2 of 70 mm Hg (Table 2). Three patients declined to have ABG drawn.

Lung volumes were normal in 15 of 16 patients. Patient 19 had a mildly increased residual volume to total lung capacity (RV/TLC) of 130%. The Dco was not available in four patients who had normal ABG values and/or pulse oximetry. The Dco was mildly reduced in patient 1 (Dco 70% of predicted) who had a normal resting ABG value and a normal exercise test.

Eleven of 16 patients who underwent exercise testing had a normal exercise capacity. Six of these displayed dystonic movements and irregular breathing patterns during exercise that contributed to their complaints of dyspnea.

Three patients gave suboptimal efforts, one of whom had dystonic movements while exercising. One patient had cardiovascular limitation, and one had ventilatory limitation that contributed to their dyspnea.

All 19 patients in group 1 underwent VF (Table 2).
Two patients had a completely normal VF. Neither had exercise testing but both had irregular FVL and flow patterns.

Seventeen patients (89%) had abnormal VF with UA dysfunction, diaphragmatic dysfunction (DD), or a combination of both. These findings on VF (Table 3) correlated with abnormalities on the FVL and dyspnea.

**Group 2: Obstructive PFT (n=3)**

Three of the 26 patients (11%) had obstructive spirometry (mean FEV₁, 60.3% ± 10.0% of predicted, and FVL (Table 2). All had a significant smoking history (>20 pack-years). Two of these patients had FVL and/or tidal volume breathing patterns with intermittent interruption of air flow during inspiration and/or expiration, sometimes accompanied by a grunt or stridor (Fig 4). Two patients had mildly decreased respiratory muscle pressures: mean Plmax, 77.0% ± 30.6% of predicted; mean PEmax, 68.3% ± 12.6% of predicted (Table 2).

The Dco was normal in two of the three patients in this group. Two patients had normal ABG values. One patient had mild hypoxemia and hypercarbia (Po₂, 72 mm Hg, and Pco₂, 47 mm Hg) (Table 2). The VF was abnormal in two of three patients. Both patients also had abnormal FVL.

All three patients underwent cycle ergometry. Patient 20 had a limited study because of poor effort. Patient 21 had dystonic movements and irregular respirations, but an otherwise normal result on the exercise test. Patient 22 had ventilatory limitation that contributed to her dyspnea.

**Group 3: Restrictive PFT (n=4)**

Four of the 26 patients (15%) had restrictive spirometry and FVL (Table 2). All four patients had FVL with intermittent interruptions of air flow during inspiration and/or expiration.

Two patients had decreased respiratory muscle pressures (mean Plmax, 40.5% ± 2.1% of predicted; mean PEmax, 33.0% ± 9.9% of predicted).

The Dco was normal in the two patients in whom it was measured. Arterial blood gas determination was not available for patient 24 and was normal in patients 23 and 26. Patient 25 had a Pco₂ of 47 mm Hg.

Lung volumes were measured in two patients, one of whom had a reduced TLC (61%).

The two patients who had an exercise test had dystonic respiratory movements. Videofluoroscopy was abnormal in all four patients (Table 2).

Overall, respiratory muscle pressure was reduced in 9 of 26 patients (34%). Patient 25 could not perform respiratory muscle pressure testing due to excessive oral movements, with inability to close his mouth securely enough around the mouthpiece.

Twenty of 24 patients (83%) had abnormal FVL. The most common abnormalities were interruptions of flow during inspiration and/or expiration, often worsened by multiple efforts and best detected from their FVL, and the marked variations of breath-to-breath volume during tidal or MVV breathing. In most patients, flow pattern variability could not be normalized even after multiple coached efforts. Occasionally, a FVL loop would not show any irregularities.

Three of four patients with normal FVL had abnormal VF. Videofluoroscopy was abnormal in 23 of 26 patients (88%): 19 patients had both UA dysfunction and DD, patient 13 had only UA dysfunction, and 3 patients had only DD.

Table 3 lists the abnormalities observed during VF. Two of three patients with normal VF had an abnormal FVL. Overall, only 1 of 26 patients had no irregularities or interruptions in the FVL and a normal VF (patient 20). Her ABG value was normal, but her exercise test was of suboptimal effort.

Table 4 summarizes the abnormalities from all 26 patients.

**DISCUSSION**

Dyspnea in dystonia appears to be associated with spasmodic contractions of the upper airways and/or the diaphragm in most patients. Since maximum efforts are necessary for optimal performance of PFT, and increased efforts tend to worsen any volitional muscular contractions causing spasms, these efforts appear to be perceived as discomfort or dyspnea. Although patients showed a desire to comply with testing, extra efforts were necessary to overcome the spasmodic muscular contractions to produce airflow which, in turn, worsened the spasms. However, interestingly, many of our patients were able to ex-

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**Table 3—Videofluoroscopic Findings**

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<td>1. Tight adhesion of true/false vocal folds</td>
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<td>2. Repeated rhythmic elevation of the uvula</td>
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<tr>
<td>3. Delay in laryngeal relaxation</td>
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<td>4. Incomplete adduction of vocal folds</td>
</tr>
<tr>
<td>5. Vocal folds adductor spasms during inspiration</td>
</tr>
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<td>6. Adductor tremor of vocal folds/tongue/uvula</td>
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<th>Diaphragmatic dysfunction</th>
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<tr>
<td>7. Poor to no hemidiaphragmatic coordination</td>
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<td>8. Spasmodic contraction of hemidiaphragm</td>
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<tr>
<td>9. Delayed relaxation of hemidiaphragm</td>
</tr>
<tr>
<td>10. Spasmodic tremor of hemidiaphragm: part of one, one, both</td>
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</table>

*Table 3 includes patients with other additional findings noted in the videofluoroscopy reports: 19 patients had both upper airway and diaphragmatic dysfunction; 4 patients had diaphragmatic dysfunction alone; 1 patient had upper airway dysfunction alone; and 3 patients had normal videofluoroscopy.

**Table 4** summarizing the abnormalities from all 26 patients.
Submaximal efforts despite the patient’s coached attempts to give a “best effort,” because such increased efforts caused excessive discomfort from muscle spasms. It is also possible that UA spasm during maximal efforts reduced the pressures recorded at the mouth that do not accurately reflect the true respiratory muscle pressures generated.

Esophageal and gastric balloon pressures were not measured. However, Dr. T. Aldrich evaluated body-box flow-resistance in patients 19 and 22 and was unable to discriminate UA from diaphragmatic source of spasms (personal communication, 1993). As a group, the patients with restrictive deficits had the lowest respiratory muscle pressures. Their evaluation excluded primary parenchymal or vascular lung abnormalities. All four patients with restrictive PFT had generalized dystonia, and three of four had severe dyspnea limiting their daily activity. When dystonic patients have restrictive PFT, reduced respiratory muscle strength could be the cause of restriction.

Exercise performance was better than expected by patients, despite dysynchrony in some. The ability to perform exercises safely, as confirmed by good gas exchange, reassured both patient and referring physician, and allowed a prescription for a reconditioning program. Whether deconditioning, resulting from limiting activity to reduce spasms, imposes an added element to dyspnea, as in patients with COPD, cannot be determined at this time. If reconditioning reduces dyspnea, then the patient’s capacity to function can be improved, and thus enhance quality of life. Efforts generated during exercise are reflexive and different from the maximal volitional responses required for PFT performance.

In most patients, ABG and/or Dco were normal, indicating that the movement disorder does not usually affect gas exchange. In the patients with mild abnormalities, the changes appear too small to account for the dyspnea. In contrast to patients with respiratory dysrhythmia due to tardive dyskinesia, respiratory alkalosis was not present in our patients. This lack of respiratory alkalosis may be due to the episodic nature of upper airway/diaphragm spasms in dystonic patients. In addition, the mild hypoxemia at rest seen in one smoker (patient 21) was reversed during cycle ergometry, indicating adequate ventilatory reserve. Minimal hypercapnia was noted in three patients: one had normal PFT, one had mild obstruction, and one had mild restriction.

The elevated Pco2 in these patients, with normal or mildly abnormal PFT, could be due to more severe UA obstruction resulting from spasm of the vocal folds. Asynchronous movements in the standing/sitting position were often reduced or abolished when reassessed in the supine posture. This positional im-

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*Only 1 of 26 patients had normal flow volume loop (FVL) and videofluoroscopy (VF).
1In group 1, only 17 patients had FVL results available. Of these, 3 had normal FVL. All three had abnormal VF.
1Muscle P refers to respiratory muscle pressure (ie, Plmax and PEmax).
1Two of three patients with normal VF had abnormal FVL.

cercise. This suggests a different pathway for effort generation in exercising as opposed to the efforts required for PFT.

It is important to directly observe patients during testing, and not discard any patient’s PFT or FVL as “poor effort” or “technically inadequate.” Each tracing should be examined for flow interruptions. In 20 of 24 patients (83%), flow patterns could not be normalized despite repeated attempts. These flow interruptions originated from spasmodic contractions of either the UA and/or the diaphragm, best seen during VF.

It was not technically possible to differentiate their respective or dominant contribution because VF could not be done simultaneously with PFT. However, auscultation at the neck and direct palpation of the chest wall and diaphragm (at the costal margins) could, at times, make differentiation possible. It also became clear during VF that greater attempts to overcome the spasms only worsened them.

Furthermore, nose breathers were found to have less dystonic movements of the diaphragm than mouth breathers. Pulmonary function testing obligates the oral route and thus heightens the dystonia. This was best observed during VF when dystonic movements increased with oral panting, and less so during quiet nasal respiration. The supine posture was associated with a reduction or cessation of spasms, which was reflected by improved speech and less dyspnea in most patients. The correlation of patients’ history with VF was remarkable: the position chosen by the patient as causing the least spasm and offering the greatest comfort was confirmed by VF. Stridor may not be audible when flow rates are low (Fig 3 A and B), but is more likely to be heard with higher flow rates. This is illustrated by a different patient (Fig 4) whose flow rate is double that of the patient in Figure 3B.

The cause or causes of reduced respiratory muscle pressures in 9 of 26 patients (34%) is unclear. It could be due to deconditioning. It could also be due to...
progression of patients’ symptoms correlated with improved spirometry and FVL. We speculate that when postural muscles are not already contracted, maximal efforts generate less spasm. The mechanism for this is unknown.

Spasmodic contractions of UA and/or diaphragm during quiet breathing and/or speaking associated with the sensation of excessive effort to generate airflow to overcome the spasms are best seen during VF. Altered speech with reduced speaking capacity (dysphonia), due to upper airway spasms, was the most frequent reason for seeking medical attention. Thus, speech impediment appears to contribute to the sensation of dyspnea in dystonic patients and is a helpful diagnostic clue to possible diaphragmatic dysfunction.

One patient (patient 22) had phrenic nerve testing with VF: phrenic nerve latency was normal, as was the diaphragmatic contractile response. This finding suggests that the peripheral nerve-muscle axis is intact, and that her dystonic movements originated from higher neural centers.

Irrespective of the etiology, the respiratory awareness described as dyspnea in patients with dystonia appears to originate from spasmodic or dysynchronous contractions of the UA and/or diaphragm.

When the dyspnea score (Table 2, graded 1 to 4) was compared with the dystonia type (focal=1, multifocal=2, generalized=3), the correlation coefficient was 0.61, suggesting that increasing dyspnea was associated with a more generalized dystonic process. However, when the dyspnea score was compared with the number of abnormal test results for each patient (eg, abnormal FEV1/FVC is one; abnormal FEV1/FVC and lung volume is two; abnormal flow volume loop, Plmax, and ABG (three, etc), a poor correlation (R=0.37) was found. Similarly, no correlation was found when the number of abnormalities was compared with dystonia type (R=0.45).

The literature is very limited regarding breathing abnormalities in dystonic patients. LaBlance (1982) in a dissertation article with emphasis on speech affectation, compared the breathing patterns of six patients (mean age, 28.65 years) with generalized dystonia to four normal control subjects (mean age, 29.21 years). None of his patients had respiratory complaints. Prior to this study, five of six patients with dystonia had undergone thalamic surgery in an attempt to improve the movement disorder. This author did not find any statistical difference between the patients and the control subjects in mean rate of respiration at rest or during speech, and did not report any consistent effect of body position (sitting, standing, or supine) on breathing patterns between the two groups. He did describe some chest/abdominal movement asynchrony in the dystonic group that improved in the supine position and worsened during speech. Respiratory pauses were described in the dystonic group, more so in the supine position. Greater variability in every aspect of the breathing pattern was seen in the dystonic group without any difference in arterial oxygen saturation (SaO2) between the two groups, or during changes in position. Arterial blood gases were not measured. Like our patients, LaBlance (1984) also noted significant differences in tidal volume during both quiet breathing and speech between the two groups: lower tidal volume and greater within-subject variability of tidal volume being noted in the dystonic patients.

Dystonic patients without respiratory complaints had differences only in breathing patterns when compared with normal subjects. These differences were influenced by position changes and speech.

Studies in patients with extrapyramidal movement disorders are few. Weiner et al (1980) described four patients with drug-induced dyskinesia who had dyspnea and chest pain secondary to the movement disorder. Three of four had normal PFT results; the fourth patient could not effectively apply her lips to the apparatus to have PFT. All four patients had irregular respiration (both in rate and depth) and involuntary grunts and gasps, similar to some of our patients. Flow volume loops were not described. Abnormal movements and breathing irregularities disappeared during sleep. The respiratory abnormalities were worse when the movement disorder worsened. Ivanovich et al (1981) described incapacitating respiratory distress in one patient with a choreiform movement disorder. This patient had mild obstructive PFT with normal Dco but with variation in respiratory rate and tidal volume. The FVL of their patient is not described, but VF of the UA showed abnormal movements of the tongue and pharynx. They did not assess the diaphragm. Like our patients, dyspnea worsened with stress or exercise, but disappeared during sleep.

Wilcox et al (1982) in evaluating the respiratory muscles in patients with tardive dyskinesia, concluded that “these patients had irregular rapid shallow breathing which is less variable during sleep, and which does not limit their exercise performance.” As reported by patients’ spouses and confirmed in all four patients who underwent sleep testing, dystonic movements usually disappeared during sleep. The improvement of respiratory muscle spasm during sleep was also described by Kuna and Awan (1985) during a sleep study of a patient with respiratory dyskinesia. The one exception (patient 26) had severe generalized dystonia. Her sleep-associated dystonic movements were reduced with a ventilatory support system (BiPAP, Respironics Inc, Murrysville, Pa).
Thus, tardive dyskinesia and dystonia appear to share the common findings of variation in tidal volume breathing, improvement by assuming a supine position, normal exercise capacity in most, and disappearance of the abnormal movements during sleep.

Dyspnea in patients with dystonia requires a thorough evaluation to exclude other pulmonary and/or cardiac causes. The variability of testing abnormalities present in the same patient, and among different patients, makes it impossible to predict which test would best demonstrate the abnormalities present in any one subject at any time. Direct observation of patients during PFT with flow interruptions in the FVL (not correctable with additional coaching) coupled with dynamic VF of both the UA and diaphragm appear to be the most effective way to diagnose respiratory system involvement by dystonia.

To our knowledge, this report is the first systematic assessment of the UA and diaphragm in dystonic patients with dyspnea. Assessing and documenting these abnormalities benefit dystonic patients by determining the origin of their symptoms, validating their complaints, and serving as a valuable aid in counseling. Testing also establishes the pathophysiology from which an objective assessment of the responses to therapy can be compared, and allows physicians a method with which to follow each patient for the rate and degree of changes over time and/or with therapy. It is hoped that by expanding our understanding of the spectrum of dystonic symptoms, new avenues will open for the study of respiratory function in a disease with an elusive etiology.

ACKNOWLEDGMENTS: The authors gratefully acknowledge Dr. Joseph Ghassibi’s help with exercise testing, and Beverly Cayetano’s assistance with manuscript preparation.

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