

Assessment of diaphragm motion using ultrasonography in a patient with facio-scapulo-humeral dystrophy

A case report

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Abstract

Rationale: Diaphragm is the main inspiratory respiratory muscle and little is known about diaphragm ultrasound in facio-scapulo-humeral muscular dystrophy, a neuromuscular disease characterized by an asymmetric skeletal muscle involvement.

Patient concerns: Diaphragm function evaluation

Diagnosis: Diaphragm muscle weakness attested by the drop of vital capacity (VC) value from sitting position (74%) to supine position (46%).

Interventions: A diaphragm ultrasound was performed in supine position, from the anterior subcostal window

Outcomes: We found an opposite side to side hemi diaphragm displacement, either during sniff maneuver or during deep inspiration maneuver, showing a cranial abnormal reduced motion of the right hemi diaphragm whereas the left hemi diaphragm moved caudally.

Lessons: Diaphragm weakness may be present with an asymmetric pattern and an opposite motion during inspiration or sniff manoeuvre in facio-scapulo-humeral muscular dystrophy. A future study with a systematic evaluation of a greater number of FSHD1 patients will be necessary to characterize this population.

Abbreviations: FSHD = facioscapulo-humeral dystrophy; MIP = maximal inspiratory pressure; SNIP = sniff nasal inspiratory pressure; VC = vital capacity.

Keywords: diaphragm weakness, FSHD1, ultrasound

1. Introduction

Facioscapulo-humeral dystrophy (FSHD) is the third most common genetic muscular dystrophy, with an estimated prevalence of 1/8000.^[1,2] This disease is characterized by an asymmetric muscular involvement, affecting initially the face and the shoulder girdle, followed by the truncal, humeral and lower extremity muscles. Among FSHD forms, the autosomal dominant FSHD1 is the most prevalent one (95%). This disease results from the loss of a subset of repeated units in the D4Z4

macrosatellite repeat array (chromosome 4q35)^[3,4] that leads to an aberrant production of DUX4, a double homeobox transcription factor with toxic gain of function. Respiration function may be involved, providing a restrictive pattern^[5-7] and a smaller D4Z4 size seems to be associated with a more severe form with an earlier onset of disease.^[8] We report here a side-to-side opposite diaphragm motion using sonography in a patient with FSHD1. Informed consent was obtained from the patient for this case report.

2. Case report

A 61 year-old female with facio-scapulo-humeral dystrophy (FSHD1) was referred to our unit for evaluation. His past medical history was pertinent for diabetes mellitus and hypertension. Initial symptoms appeared at the age of 21 years with shoulder girdle weakness followed by difficulty walking. Clinical examination disclosed winged scapulae, weakness affecting the shoulder girdle, the facial, the trunk associated with a steppage gait. Cardiac function was normal with a left ventricular ejection fraction at 75%. Vital capacity (VC) value decreased from sitting position (74%) to supine position (46%), which attests diaphragm muscle impairment. In addition, sniff nasal inspiratory pressure (SNIP) was significantly decreased (17 cmH₂O) as well as maximal inspiratory pressure (MIP at 12 cmH₂O), related to a failure of strength's diaphragm muscle. Diaphragm ultrasound (Vivid E9, GE Medical Systems) was performed in

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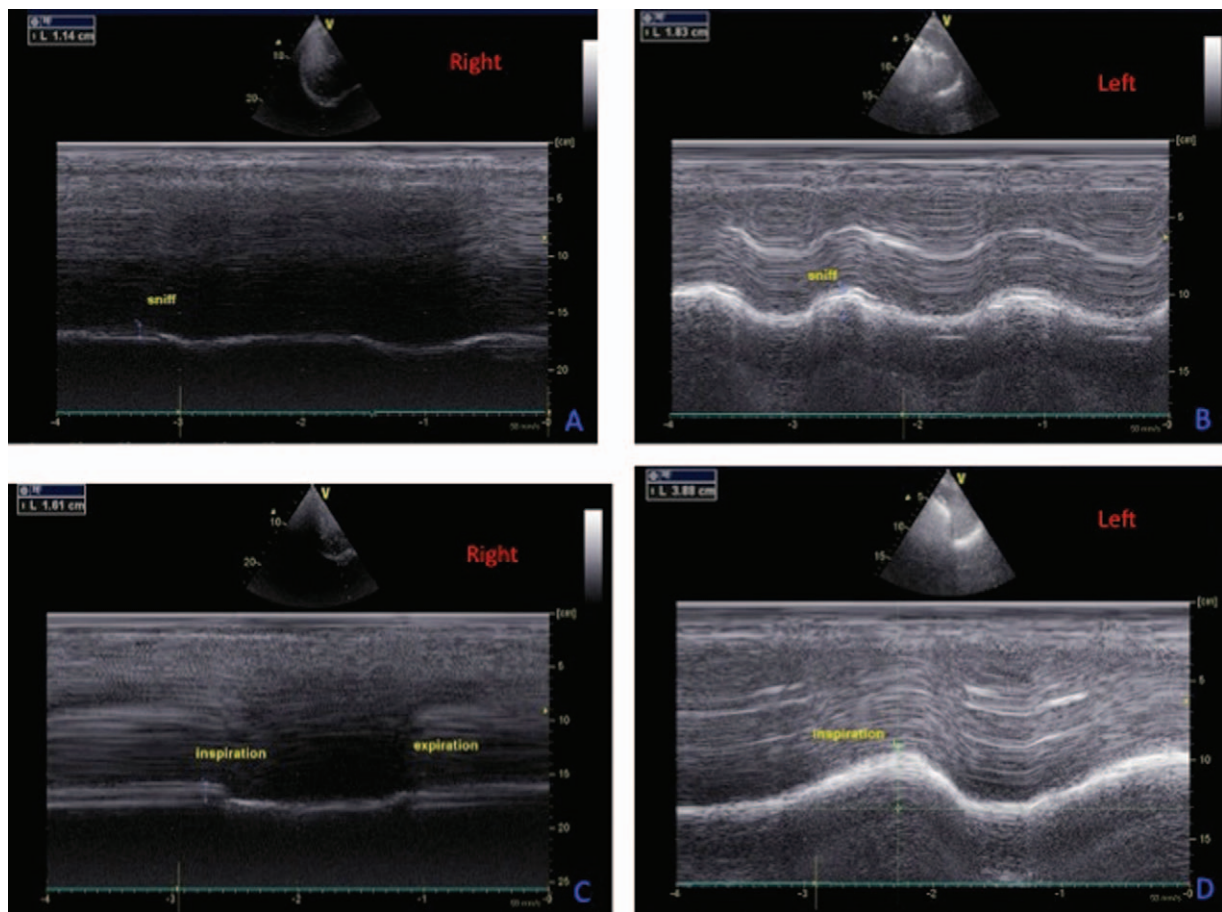


Figure 1. Diaphragm ultrasound imaging in FSHD1. The right hemidiaphragm moves cranially in an opposite direction during deep inspiration (C) or Sniff maneuver (A) in comparison with the normal caudal movement of the left hemidiaphragm during deep inspiration (D) or Sniff maneuver (B). FSHD1 = facio-scapulo-humeral dystrophy.

supine position (45°), using a sector probe M5Sc-D (1.5–4.6 MHz), from the anterior subcostal window. The imaging exam disclosed an opposite side to side hemidiaphragm displacement, either during sniff maneuver or deep inspiration maneuver, with a cranial abnormal reduced motion of the right hemidiaphragm whereas the left hemidiaphragm moved caudally during inspiration or sniff maneuver (Fig. 1).

3. Discussion

Respiratory insufficiency is a frequent cause of morbidity and mortality in neuromuscular disorders. In FSHD1, the typical clinical pattern includes asymmetric skeletal muscle impairment, with atrophy and muscle fatty replacement of trapezius, teres major and serratus anterior.^[9] Wheelchair is necessary in 20% of patients often after the 5th decade.^[2] Respiratory function is not systematically involved, and a restrictive respiratory involvement was found in 9.8% of the largest published cohort with genetic confirmation of FSHD.^[7] Only 1% of patients needed home mechanical ventilation in the Dutch FSHD population.^[10] The respiratory insufficiency is particularly frequent in the infantile form and in patients with small D4Z4 repeats sizes.^[11] Diaphragm is the main inspiratory respiratory muscle and little is known about diaphragm function in this disease. Classical pulmonary function tests assess global respiratory muscles function, and a decline in VC greater than 20% from the sitting to the supine position is

interpreted as an indirect sign of diaphragm weakness.^[12] One study assessed global respiratory muscles and diaphragm using transdiaphragmatic pressures in FSHD patients^[13] and reported a weakness affecting the expiratory muscles rather than the inspiratory muscles. Indeed, in FSHD, abdominal muscles—that are the main expiratory muscles—seem to be early affected, in association with protuberant abdomen and lumbar lordosis.^[10,14] The *Beevor sign* may be present, due to selective weakness of the lower rectus abdominis. Contrarily to the study by Stübgen and Schultz,^[13] that did not find any alteration of diaphragm function in FSHD using transdiaphragmatic pressures measurement, we found an asymmetric and opposite diaphragm motion in FSHD1 using sonography. This difference can be explained by the fact that transdiaphragmatic pressures may remain in normal range in patients with asymmetric diaphragm failure, since transdiaphragmatic pressures measure the global diaphragmatic function as the pressure difference between the pleural pressure (oesophageal) and the gastric pressure. On the contrary, the use of ultrasound allows detecting asymmetric diaphragm weakness.^[15,16] For the diagnosis of diaphragm paralysis, chest X ray may have a significant sensibility but a lower specificity. Fluoroscopy was traditionally used for the diagnosis of diaphragm paralysis, showing a paradoxical motion during Sniff manoeuvre but this technique is currently outdated. Diaphragmatic muscle strength may be explored by the measurement of maximal static inspiratory pressure and sniff nasal inspiratory pressure.^[17] These noninvasive

methods showed an excellent correlation ($0.99 + -0.01, p < 0.001$) with sniff oesophageal pressure.^[17] Ultrasound imaging is a noninvasive approach for diaphragm analysis and can be coupled with sniff manoeuvre. In our case, the weakness of diaphragm was confirmed by sniff, MIP, delta VC (decrease of VC from sitting to supine position) measurements and ultrasound imaging. Our results support the fact that diaphragm muscle may be impaired in FSHD1 with an asymmetrical distribution, and the disease seems thus to not affect only the expiratory respiratory muscles, as previously described. The diaphragm involvement was asymmetric in our case, likewise the asymmetric skeletal muscle involvement, a hallmark of FSHD1. This was not yet reported in the literature, except for one study that reported a bilateral diaphragm paralysis in a patient with FSHD.^[18] Diaphragmatic ultrasound could be a sensitive monitoring tool in addition to functional respiratory tests. Our study should be considered as exploratory, since it disclosed some limitations that need to be addressed: we report the result of only one patient and we do not perform oesophageal pressure measurement.

Author contributions

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