

# Prevalence and impact on quality of life of gastrointestinal and genitourinary symptoms in facioscapulohumeral muscular dystrophy

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## Objective

To explore the prevalence of gastrointestinal (GI) and genitourinary (GU) symptoms and their impact on quality of life in people with facioscapulohumeral muscular dystrophy (FSHD) compared to healthy household controls

## Background

Anecdotally, patients with FSHD describe GI and GU symptoms. Similar symptoms are reported in patients with other muscular dystrophies<sup>1,2,6,7</sup> but the prevalence is unknown in FSHD<sup>4,5</sup>

## Design and Methods

- Survey based on validated symptom scales and clinical criteria, slightly modified from survey used in similar study<sup>3</sup> in limb girdle muscular dystrophy
- Survey link distributed by email to all FSHD Society patient contacts (n=3507)
- All respondents  $\geq 18$  years old were analyzed (n=702; 652 with FSHD; 19% response rate)
- Responses included a range of frequency options (“never” to “>5x/week” or “never” to “every time”)
- In Tables 1 and 4 any positive (non-“never”) response is considered “yes” for display purposes though analysis accounted for the full range of responses
- All useable data were included – N varies by question as not all respondents answered every question

## Demographics

	FSHD	Controls
<b>Age (years)</b>		
Median [IQR]	56 [42-66]	56 [52-68]
Range	18-90	26-85
<b>Sex</b>		
Male	44.3%	38%
Female	55.2%	62%
Self or family member with genetically-confirmed diagnosis		611/647 94.4%
Able to walk without using a cane, walker, wheelchair or scooter		383/648 59.1%
Require respiratory support		90/646 13.9%

## Results

**Table 1.** Symptom frequency, respondents with FSHD vs healthy household controls

Swallowing symptoms (from last month), count (%)	FSHD	Control	p-value*
<b>Difficulty swallowing food</b>	(n=647)	(n=48)	<b>0.005</b>
No	368 (57)	36 (75)	
Yes	279 (43)	12 (25)	
<b>Modify how you ate based on swallowing difficulties</b>	164/645 (25)	6/48 (12)	<b>0.045<sup>^</sup></b>
<b>Gastrointestinal symptoms (from last month), count (%)</b>	<b>FSHD</b>	<b>Control</b>	<b>p-value*</b>
<b>Had abdominal pain</b>	(n=625)	(n=45)	<b>0.014</b>
No	237 (38)	24 (53)	
Yes	388 (62)	21 (47)	
<b>Problems with constipation</b>	(n=634)	(n=45)	<b>0.026</b>
No	227 (36)	22 (49)	
Yes	407 (64)	23 (51)	
<b>Unable to hold bowel movement until could get to restroom</b>	(n=625)	(n=43)	<b>0.0002</b>
No	397 (64)	39 (91)	
Yes	228 (36)	4 (9)	
<b>Abdominal pain caused me [degree of discomfort]</b>	(n=562)	(n=40)	<b>0.0009</b>
No	169 (30)	20 (50)	
Yes	393 (70)	20 (50)	
<b>Urinary symptoms (from last month), count (%)</b>	<b>FSHD</b>	<b>Control</b>	<b>p-value*</b>
<b>Urinate again within 15 minutes</b>	(n=622)	(n=42)	<b>0.002</b>
No	332 (53)	33 (79)	
Yes	290 (47)	9 (21)	
<b>Had episode of wetting self in the daytime</b>	(n=626)	(n=43)	<b>0.027</b>
No	401 (64)	35 (81)	
Yes	225 (36)	8 (19)	

**Table 2.** Symptoms with no difference in prevalence, FSHD vs household controls

Swallowing Symptoms
Difficulty swallowing liquids
Coughing or choking associated with eating or drinking
Felt like food “got caught” on the way down
Heartburn or reflux
Modified what you ate based on swallowing difficulties
Gastrointestinal Symptoms
Diarrhea
Painful bowel movements
Frequency of bowel movements
Urological Symptoms
Push or wait to start passing urine
Urine stream stopped and started multiple times or seemed weak
Felt bladder was not empty after urinating
Dysuria
Nocturnal enuresis

There was **no difference** between groups in the use of medications for the reported symptoms.

**Table 3.** Symptom frequency >1x/week or >50% of the time, FSHD vs household controls

	FSHD	Control	p-value
<b>Symptoms from last month, count/n (%)</b>	(n=652)	(n=50)	
<b>Swallowing Symptoms (&gt;1x/week)</b>			
Difficulty swallowing food	105/647 (16)	2/48 (4)	0.026
Felt like food “got caught” on the way down	113/642 (18)	2/48 (4)	0.016
<b>Gastrointestinal Symptoms (&gt;1x/week)</b>			
Had abdominal pain	145/625 (23)	4/45 (9)	0.026
Problems with constipation	182/634 (29)	6/45 (13)	0.026
Painful bowel movements	83/620 (13)	1/44 (2)	0.032

To account for the possibility that the findings in Table 1 were driven by infrequent symptoms (e.g. <1x/month), Table 3 includes analysis of only those symptoms occurring at higher frequencies and more likely to impair quality of life.

**Table 4.** Symptom frequency, FSHD requiring no support vs FSHD requiring respiratory support

Swallowing symptoms (from last month), count (%)	No Support	Respiratory	p-value*
	(n=356)	(n=90)	
Difficulty swallowing food	128/355 (36)	56/89 (63)	<b>&lt;0.0001</b>
Difficulty swallowing liquids	85/355 (24)	47/89 (53)	<b>&lt;0.0001</b>
Coughing/choking associated with eating/drinking	157/353 (44)	61/89 (69)	<b>&lt;0.0001</b>
Felt like food “got caught” on the way down	145/351 (41)	64/89 (72)	<b>&lt;0.0001</b>
Had heartburn or reflux	170/352 (48)	58/90 (64)	<b>0.0003</b>
Modify how you ate based on swallowing difficulties	58/353 (16)	42/89 (47)	<b>&lt;0.0001</b>
Modify what you ate based on swallowing difficulties	28/353 (8)	27/89 (30)	<b>&lt;0.0001</b>
<b>Gastrointestinal symptoms (from last month), count (%)</b>			
Had abdominal pain	194/340 (57)	71/90 (79)	<b>0.0003</b>
Problems with constipation	198/344 (58)	68/88 (77)	<b>0.0007</b>
Painful bowel movements	141/338 (42)	50/84 (60)	<b>0.013</b>
Unable to hold bowel movement until get to restroom	82/338 (24)	52/87 (60)	<b>&lt;0.0001</b>
Abdominal pain caused me [severity of discomfort]	195/307 (64)	70/82 (85)	<b>&lt;0.0001</b>
<b>Urinary symptoms (from last month), count (%)</b>			
Push or wait to start passing urine	92/341 (34)	38/85 (45)	<b>0.0003</b>
Felt bladder was not empty after urinating	155/338 (46)	51/86 (59)	<b>0.026</b>
Hurt when urinating	21/336 (6)	18/87 (21)	<b>0.0001</b>
With a strong urge to urinate, need to do something to stop from wetting	143/338 (42)	70/86 (81)	<b>&lt;0.0001</b>
Had episode of wetting self in the daytime	81/338 (24)	43/86 (50)	<b>&lt;0.0001</b>
Had episode of bed wetting	16/336 (5)	15/86 (17)	<b>0.0001</b>

\*Data was collected using scale rating, but categories are collapsed here for presentation. P values calculated based on original categories

For the analysis in Table 4, we used the need for respiratory support as an indicator of more advanced disease progression

## Effect on Quality of Life

- Respondents with FSHD were significantly more likely than controls to report that symptoms in each of the symptom categories negatively impacted their quality of life
  - Swallowing (p=0.0009)
  - Bowel (p<0.0001)
  - Urinary (p<0.0001)

## Conclusions

- GI and GU symptoms occur more frequently in people with FSHD compared to controls
- Symptom frequency appears to increase with disease progression
- Contributing factors could include impaired mobility or disease-related muscle involvement
- GI and GU symptoms impair quality of life for some people with FSHD and this may improve with improved symptom management
- Further study of causes and treatments is indicated

## References

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