

# COGNITIVE AND PSYCHIATRIC ALTERATIONS IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY: A CASE REPORT

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## Introduction

Central nervous system disturbances in facioscapulohumeral muscular dystrophy (FSHD) were thought to be minimal in contrast with other muscular disorders. However, recent studies detected brain atrophy, alterations of neuronal networks, reduced cognitive functions in patients with large gene deletions. In addition, it is becoming increasingly evident that non-muscular symptoms do not depend from the contraction size of D4Z4 allele and they are not proportional to muscle impairment. Few articles studied the association between psychiatric disorders and FSHD: personality, behavioural disturbances, depression, phobias were detected; a single association was made with schizophrenia.

## Methods

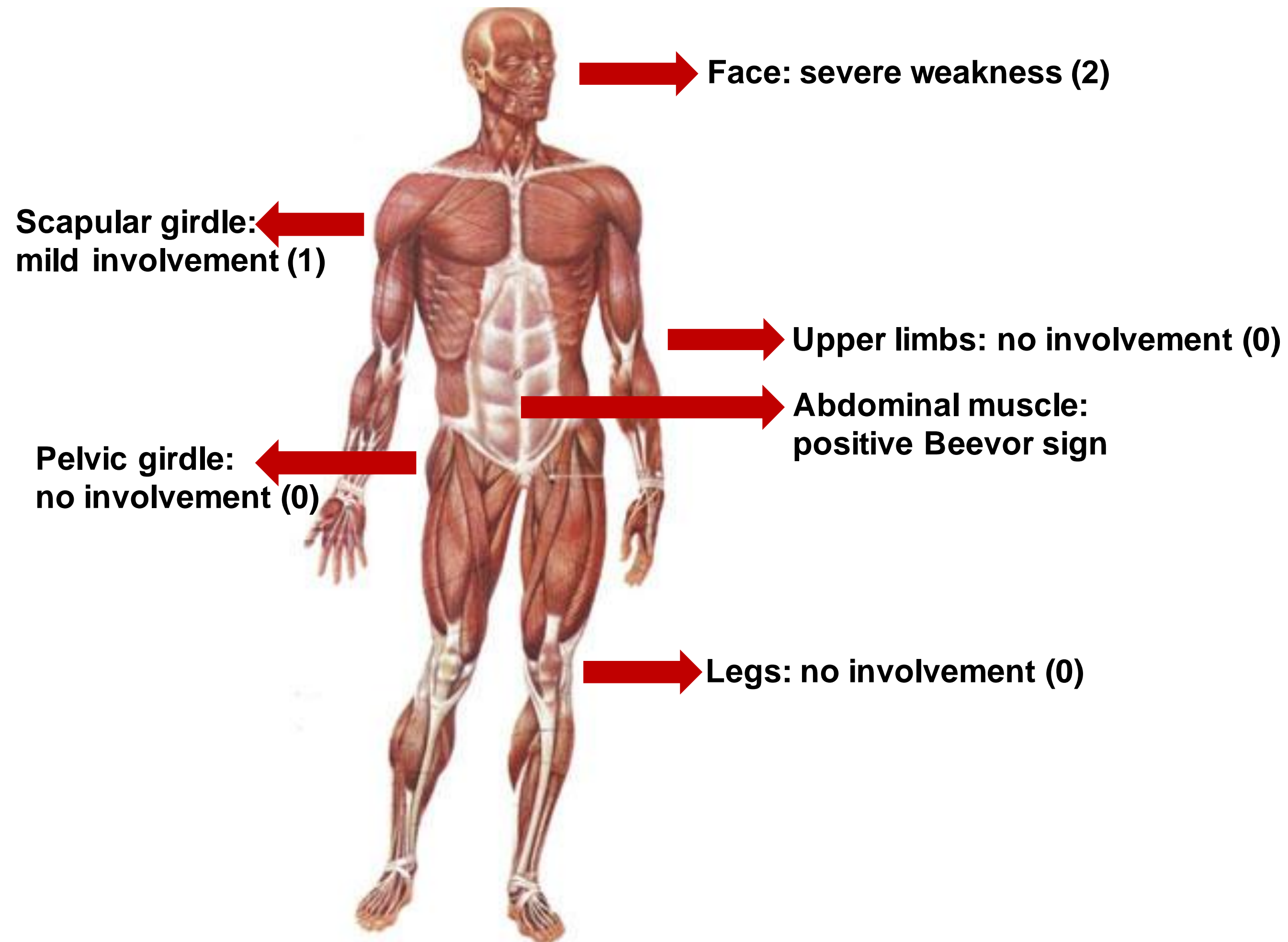
We describe a 30 years-old woman with a one-year history of facial and shoulder girdle muscles weakness, exacerbated after the first pregnancy (positive family history for FSHD). Since the first examination, behavioural disturbances and fatuous attitude were evident: the patient was irritable and aggressive against doctors and husband. A routine psychological interview pointed out compulsive and obsessive rituals focused on cleanliness and avoiding illness towards herself and her child.

## Results

The genetic testing confirmed the diagnosis of FSHD1 (fragment of 25 kb). The neuropsychological evaluation showed impairment of executive functions, reasoning and visuo-praxic skills. The psychiatric evaluation concluded for adjustment disorder with obsessive compulsive disorder (OCD). A pharmacological therapy with sertraline was started with improvement of behavioural symptoms, quality of life and disease disability perception.

## Conclusion

To the best of our knowledge, this is the first report of association between FSHD and OCD. Furthermore, this case confirms that additional factors should be analysed to explain extra-muscular involvement in FSHD.



The FSHD clinical evaluation of our patient: FSHD score 4

## Neuropsychological assessment

Test	R.S.	C.S.	E.S.
<b>MMSE</b> (n.v. ≥ 24)	27	25,75	
<b>Verbal Functioning</b> Phonemic Fluency	31	25,5	2
<b>Memory ST</b> Digit Span Reverse Digit Span	5 2	4,5	2
<b>Memory LT</b> Short Story (A. Pesenti) Rey Figure Recall	14,5 11	11,5 3,75	2 0*
<b>Praxis</b> Rey Figure Copy	32	29,5	1
<b>Attention/Executive Functions</b> Attentive Matrices Trail Making Test A Trail Making Test B Trail Making B-A Stroop Time (n.v. ≤ 36.92) Errors (n.v. ≤ 4.24) Cognitive Estimation (n.v. ≤ 18) Errors (n.v. ≤ 4) Clock Drawing Test (n.v. ≥ 8)	42 60'' 150'' 90 17 3 25 5 7/10	33,75 70 184 114 28 4,5 24,2	1 1 1 1 2 0* Insufficient Insufficient Insufficient
<b>Mood</b> STAI-Y BDI-II	<b>Not Reliable</b> <b>Not Reliable</b>		
<b>Reasoning</b> Colored Progressive Matrices	10	7,5	0*

The neuropsychological assessment shows abnormal executive functions, logic and abstract reasoning and praxis. Only language and memory are spared. MMSE (Mini Mental State Examination); ST (short term); LT (long term); STAI-Y (state-trait anxiety inventory-Y); BDI-II (Beck depression Inventory-II); RS (raw score); CS (corrected score); ES (equivalent score); n.v. (normal values).

## Neuropsychiatric inventory (NPI)

	BEFORE therapy		AFTER therapy	
	Frequency	Severity	Frequency	Severity
<b>DELUSIONS (jealousy)</b>	3	2	2	1
<b>HALLUCINATIONS (auditory)</b>	2	1	NO	NO
<b>AGITATION/AGGRESSION</b>	4	3	2	2
<b>DEPRESSION/DYSPHORIA</b>	4	3	2	1
<b>ANXIETY</b>	4	3	3	1
<b>ELATION/EUPHORIA</b>	1	1	1	1
<b>APATHY/INDIFFERENCE</b>	4	3	2	2
<b>DISINHIBITION</b>	3	3	2	1
<b>ABERRANT MOTOR ACTIVITY</b>	NO	NO	NO	NO

The NPI shows a marked improvement of the OCD, anxiety, agitation, delusions to a lesser extent. There is also a reduced level of insight and empathy. Frequency: 0 (never); 1 (rarely); 2 (sometimes); 3 (frequently); 4 (very frequently). Severity: 1 (mild); 2 (moderate); 3 (severe).