

Table 5: Prevalence and relative importance of specific symptoms in FSHD

Question	Number of Responses	Number Affected	Percent Affected	Mean Response	Population Impact score
Difficulty getting places without handicap accessibility	152	114	75	2.65	1.99
The requirement for increased planning	151	121	80.1	2.04	1.63
Difficulty playing sports	146	133	91.1	3.23	2.94
Unable to travel or tour	151	117	77.5	2.26	1.75
Increased time to complete activity	152	133	87.5	2.01	1.76
Difficulty getting in and out of vehicles	151	122	80.8	2.12	1.72
Difficulty doing yard work	148	132	89.2	2.66	2.37
Limited sexual positioning	135	102	75.6	2.37	1.79
Difficulty operating a motor vehicle	146	80	54.8	1.81	0.99
Difficulty maintaining a home	152	123	80.9	1.98	1.61
Difficulty cleaning a home	152	123	80.9	2.24	1.82
Impaired safety because of illness	150	104	69.3	1.87	1.29

Trouble getting out of a tub or shower	150	115	76.7	2.03	1.56
Having to modify a house to accommodate for disability	150	96	64	2.2	1.41
Problems doing the laundry	149	104	69.8	1.83	1.28
Impaired dancing	142	115	81	2.58	2.09
Worsening golf game	120	73	60.8	2.88	1.75
Impaired ability to exercise	146	138	94.5	2.71	2.56
Difficulty bathing or taking a shower	152	115	75.7	1.87	1.41
Trouble riding a bike	140	107	76.4	2.88	2.2
Unable to snow ski	133	100	75.2	2.86	2.15
Difficulty vacuuming	150	112	74.7	2.09	1.56
Difficulty painting	140	111	79.3	2.29	1.81
Difficulty changing a light bulb	148	118	79.7	2.35	1.87
Inability to "go out"	148	89	60.1	1.98	1.19
Impaired sexual function	140	89	63.6	1.93	1.23
Difficulty maintaining personal hygiene	151	71	47	1.54	0.72
Problems using buttons and zippers	152	91	59.9	1.4	0.84
Problems preparing food	152	97	63.8	1.7	1.09

Taking longer to do household chores	152	125	82.2	2.08	1.71
Difficulty washing dishes	150	92	61.3	1.77	1.09
Difficulty holding a milk gallon	151	110	72.8	2.13	1.55
Difficulty walking dogs	130	84	64.6	2.45	1.58
Difficulty getting off of a toilet	151	113	74.8	2.18	1.63
Problems washing windows	141	109	77.3	2.37	1.83
Decreased spontaneity of activities	147	119	81	2.29	1.85
Unable to mow the lawn	141	100	70.9	2.62	1.86
Not able to hike	145	127	87.6	2.61	2.29
Trouble with going up step ladders	145	127	87.6	2.68	2.34
Difficulty scrubbing surfaces	147	118	80.3	2.14	1.71
Difficulty with mopping	148	112	75.7	2.1	1.59
Not able to dust	147	97	66	1.69	1.12
Not able to play tennis	136	102	75	2.72	2.04
Difficulty lifting weights	141	124	87.9	2.38	2.09
Unable to get on amusement park rides	141	102	72.3	2.34	1.69
Unable to go to the beach	143	96	67.1	2.63	1.76
Unable to turn on a light with a pull cord	150	66	44	1.7	0.75

Difficulty digging with a shovel	143	112	78.3	2.17	1.7
Problems shaving	140	76	54.3	1.38	0.75
Difficulty putting on shoes	149	96	64.4	1.27	0.82
Difficulty putting on a tie	134	64	47.8	1.58	0.75
Difficulty staying active	147	111	75.5	2.25	1.7
Impaired ability to read	147	40	27.2	1.03	0.28
Difficulty swimming	138	103	74.6	2.13	1.59
Difficulty cutting meat	151	80	53	1.56	0.83
Difficulty using a hammer or other tool	147	113	76.9	1.87	1.44
Inability to keep appointments due to physical reasons	150	52	34.7	1.37	0.47
Visual loss	151	57	37.7	1	0.38
Difficulty painting	142	107	75.4	2.07	1.56
Physical fatigue	152	141	92.8	2.25	2.09
Poor endurance	149	137	91.9	2.34	2.15
Prolonged recovery time after exercise	146	124	84.9	2.14	1.82
Decreased energy	150	135	90	2.24	2.02
Decreased leg energy (stamina)	151	144	95.4	2.65	2.53
Tired muscles	151	143	94.7	2.57	2.43
Decreased activity level	152	139	91.4	2.6	2.38

Excessive sleep requirements	151	105	69.5	1.89	1.31
Breathing difficulties	152	76	50	1.67	0.84
Impaired sleep	151	93	61.6	1.92	1.18
Daytime sleepiness	151	103	68.2	1.3	0.89
Weak trunk (core) muscles	150	136	90.7	2.51	2.27
Back weakness	149	133	89.3	2.36	2.11
Difficulty bending down	149	128	85.9	2.45	2.1
Difficulty rolling over in bed	148	119	80.4	2.24	1.8
Difficulty sitting up	150	122	81.3	2.22	1.81
The need for a back brace	144	40	27.8	1.85	0.51
Weak chest muscles	147	100	68	1.86	1.27
Inability to get out of bed due to weakness	149	69	46.3	1.91	0.89
Neck weakness	149	99	66.4	1.52	1.01
Abdominal muscle weakness	150	133	88.7	2.1	1.86
Inability to do things previously done	150	143	95.3	2.76	2.63
Lack of ability to participate in fun activities	149	136	91.3	2.47	2.26
Negative perception by others	149	87	58.4	1.52	0.89
Lack of understanding by family, friends, or co-workers	148	69	46.6	1.32	0.61

Need to hide ones condition from friends	148	55	37.2	1.38	0.51
Lack of available information about the disease	148	52	35.1	1.5	0.53
Discrimination at work	136	28	20.6	1.79	0.37
Perceived burden to family members	149	72	48.3	1.81	0.87
Not comfortable in social environment	149	92	61.7	1.5	0.93
Unable to be in crowds	147	77	52.4	1.56	0.82
Dissatisfaction with social interactions	148	79	53.4	1.57	0.84
Lack of available information about exercise options	146	78	53.4	1.77	0.95
Increased family stress	148	88	59.5	1.57	0.93
Decreased sexual contacts	132	73	55.3	1.93	1.07
Divorce secondary to family stress	127	12	9.4	1.92	0.18
Perception of being lazy	145	59	40.7	1.32	0.54
Being perceived as a drain on society	145	27	18.6	1.3	0.24
Decreased acceptance by others	147	50	34	1.12	0.38

Being perceived by others as drunk	146	21	14.4	1.05	0.15
Memory deficits	150	67	44.7	1.07	0.48
Problems concentrating	150	64	42.7	1.14	0.49
Problems reading	151	50	33.1	1.06	0.35
The reliance on family members	151	122	80.8	1.93	1.56
Limitations physically on what one can do	152	146	96.1	2.68	2.58
Having to rely on strangers	150	100	66.7	1.47	0.98
Lack of job because of disability	145	51	35.2	2.9	1.02
Limited activities secondary to weakness	144	119	82.6	2.26	1.86
Issues at work	132	45	34.1	1.61	0.55
Difficulty interacting with family	151	50	33.1	1.3	0.43
Decreased independence	150	117	78	2.32	1.81
Impaired impression on others	145	81	55.9	1.47	0.82
Disease affecting choice of where to live	150	86	57.3	2.31	1.33
Inability to keep pace with friends while walking	145	117	80.7	2.48	2
Inability to lift children	144	121	84	2.45	2.06

Reliance on friends	149	108	72.5	1.7	1.23
Impaired social interactions	151	102	67.5	1.72	1.16
Difficulty interacting with friends	149	80	53.7	1.25	0.67
Need for reproductive intervention to have unaffected children	132	25	18.9	2.32	0.44
Difficulty with relationships	145	56	38.6	1.59	0.61
Impaired dating	133	37	27.8	2	0.56
Inability to care for family members	145	91	62.8	1.99	1.25
Lack of perfection at a job	137	53	38.7	1.53	0.59
Disability affects choice of career	140	69	49.3	2.59	1.28
The inability to hold a job	139	32	23	3	0.69
The avoidance of social situations	149	86	57.7	1.67	0.97
Inability to provide financially for family	141	56	39.7	2	0.79
Reliance on co-workers	133	47	35.3	1.34	0.47
Decreased ability to see friends	151	85	56.3	1.65	0.93
Alcohol use	145	33	22.8	0.94	0.21

The inability to hold health or long term insurance	140	40	28.6	2.25	0.64
Inability to take care of oneself	149	83	55.7	1.76	0.98
Marijuana or other drug use	142	6	4.2	0.5	0.02
Unable to babysit due to disability	142	54	38	1.94	0.74
Financial issues associated with disability	145	66	45.5	1.74	0.79
Inability to drive a car	142	37	26.1	2.73	0.71
The change from a leader to a follower	144	60	41.7	1.68	0.7
Impaired walking	174	158	90.8	2.8	2.54
Difficulty with stairs	174	161	92.5	2.99	2.77
Use of assistant devices for mobility (wheelchair, etc.)	171	109	63.7	2.53	1.61
Falls	173	135	78	1.85	1.45
Leg weakness	173	163	94.2	2.53	2.38
Whole body weakness	173	158	91.3	2.16	1.97
Difficulty with balance	173	151	87.3	2.25	1.96
Difficulty walking long distances	171	156	91.2	3.1	2.82
Inability to run	171	158	92.4	2.94	2.72

Difficulty rising from a seated position	174	158	90.8	2.41	2.19
Foot drop	169	140	82.8	2.31	1.91
Difficulty getting up from a lying position	174	159	91.4	2.01	1.84
Difficulty walking up hills or inclines	171	156	91.2	2.78	2.53
Trouble getting around	170	142	83.5	2.32	1.94
Difficulty with rough ground	171	151	88.3	2.56	2.26
Difficulty standing	171	143	83.6	2.21	1.85
Decreased pace of walking	169	153	90.5	2.54	2.3
Difficulty walking on ice	165	138	83.6	2.95	2.47
Leg wobbling	170	122	71.8	2.13	1.53
Tripping	170	147	86.5	2.06	1.79
Ankle weakness	171	126	73.7	1.96	1.44
Difficulty lifting legs	171	143	83.6	2.17	1.82
Impaired flexibility	170	141	82.9	1.96	1.63
Lack of job secondary to leg weakness	139	66	47.5	2.11	1
Knee weakness	172	124	72.1	1.79	1.29
Difficulty picking things up with your fingers	173	93	53.8	1.44	0.77
Hand weakness	174	105	60.3	1.62	0.98

Difficulty doing things with your hands	174	106	60.9	1.49	0.91
Impaired ability to open doors or drawers	173	91	52.6	1.3	0.68
Dropping objects	174	115	66.1	1.51	1
Difficulty opening jars or bottles	174	131	75.3	1.66	1.25
Worry about how others perceive you	174	138	79.3	1.49	1.18
Impaired body image	174	144	82.8	1.69	1.4
Impaired body image due to gait	163	125	76.7	1.71	1.31
Impaired body image due to abdomen protrusion	171	134	78.4	1.9	1.49
Feeling different from other people	173	136	78.6	1.63	1.28
Impaired image due to shoulder winging	173	135	78	1.52	1.18
Self conscious of muscle loss	174	152	87.4	1.7	1.48
Impaired self image	171	137	80.1	1.61	1.29
Embarrassment over Popeye arms	165	68	41.2	1.32	0.55

Embarrassment in wearing a bathing suit	171	132	77.2	1.83	1.42
Modification of diet to increase muscle size	164	49	29.9	1.02	0.3
Difficulty lifting objects	174	167	96	2.53	2.43
Difficulty reaching objects overhead	173	166	96	2.7	2.6
Difficulty brushing or washing hair	172	144	83.7	1.94	1.62
Arm weakness	174	158	90.8	2.41	2.18
Shoulder weakness	174	167	96	2.35	2.25
Reduced arm and shoulder range of motion	172	164	95.3	2.34	2.23
Difficulty putting away dishes overhead	174	161	92.5	2.4	2.22
Difficulty carrying a load	174	164	94.3	2.66	2.51
Difficulty brushing teeth	173	97	56.1	1.38	0.77
Difficulty lifting laundry baskets	169	140	82.8	2.25	1.86
Trouble hanging up a coat	171	132	77.2	1.88	1.45
Unable to take pictures	168	101	60.1	1.63	0.98
Difficulty reaching items on the back of a shelf or fridge	171	139	81.3	2.1	1.71
Difficulty lifting kids	166	145	87.3	2.76	2.41

Difficulty lifting a golf bag	158	92	58.2	2.51	1.46
Problems swallowing	173	73	42.2	1.05	0.45
Inability to eat due to facial weakness	173	32	18.5	0.97	0.18
Drooling	174	63	36.2	0.65	0.24
Difficulty using a straw	174	62	35.6	0.87	0.31
Modification of diet due to choking	173	25	14.5	0.92	0.13
A change to softer foods	170	21	12.4	1.14	0.14
Fear of progression of disease	169	158	93.5	1.93	1.8
Burden of thinking about the future	169	149	88.2	2.01	1.77
Depression	169	120	71	1.51	1.07
Damaged pride with loss of independence	168	127	75.6	1.8	1.36
Constant need to adapt to disability	168	139	82.7	1.93	1.6
Embarrassment of disability	168	126	75	1.56	1.17
Frustration	168	154	91.7	2	1.83
Awareness of the disease getting worse	169	159	94.1	2.03	1.91
Fear of passing on the disease	163	105	64.4	2.32	1.5
Fear of injury with activity	168	136	81	2.01	1.63

Anxiety	167	124	74.3	1.56	1.16
Fear of falling	166	138	83.1	2.12	1.77
Moodiness	166	106	63.9	1.45	0.93
Anger	167	102	61.1	1.16	0.71
Fear	167	112	67.1	1.52	1.02
Guilt of passing the disease on to others	163	85	52.1	2.06	1.07
Envy of people without a disability	167	116	69.5	1.34	0.93
Changed perspective on how to live life	162	136	84	1.82	1.53
Difficulty letting people get close emotionally	166	81	48.8	1.75	0.86
Sadness	169	121	71.6	1.26	0.91
Isolation	168	87	51.8	1.47	0.76
Feeling of losing control	167	111	66.5	1.59	1.06
Increased stress	168	127	75.6	1.57	1.18
Embarrassment while talking in public	165	64	38.8	1.39	0.54
Loss of physical intimacy	165	91	55.2	2.07	1.14
Symptoms mistaken as another disease by others	164	105	64	1.39	0.89
Fear of work finding out	154	31	20.1	1.94	0.39

Grief	167	84	50.3	1.06	0.53
Self pity	166	81	48.8	0.9	0.44
Fear of choking	168	55	32.7	1.29	0.42
Hypervigilance	150	62	41.3	1.4	0.58
Feeling of being overwhelmed	166	90	54.2	1.34	0.73
Regret of getting tested and knowing status	165	19	11.5	1.11	0.13
Shyness	165	67	40.6	1.21	0.49
Forced to disclose disease due to disability	166	61	36.7	1.57	0.58
Frustration with the decision to test children genetically	162	38	23.5	1.08	0.25
Feeling older than age	166	97	58.4	1.88	1.1
Obsession about the disease	169	68	40.2	1.26	0.51
Hopelessness	167	68	40.7	1.29	0.53
Fear of death	167	50	29.9	1	0.3
Fear to tell offspring that they have the disease	162	37	22.8	1.54	0.35
A feeling of being disconnected	167	76	45.5	1.39	0.63
Impaired facial expression	173	127	73.4	1.1	0.81
Impaired speech	174	75	43.1	0.87	0.37
Inability to smile	174	89	51.1	1.4	0.72

Facial weakness	173	132	76.3	1.07	0.82
Hearing difficulties	173	80	46.2	1.29	0.6
Difficulty communicating with others	174	48	27.6	1.02	0.28
Poor pronunciation	172	71	41.3	0.97	0.4
Not wanting to talk because of voice	174	24	13.8	1	0.14
Having to repeat oneself often	174	45	25.9	0.93	0.24
Can not close eyes	173	81	46.8	1.09	0.51
Difficulty writing	173	67	38.7	1.12	0.43
Impaired ability to physically demonstrate tasks	171	109	63.7	1.6	1.02
Cant raise hands	172	105	61	1.93	1.18
Difficulty interpreting non-verbal communication	172	30	17.4	0.9	0.16
Cant whistle	172	130	75.6	1.03	0.78
Quiet voice	172	54	31.4	0.94	0.3
Impaired ability to change the inflection of ones voice	172	26	15.1	1.23	0.19
Can not wink	171	57	33.3	0.8	0.27
Back pain	174	147	84.5	1.93	1.63
Diffuse pain	156	101	64.7	1.62	1.05
Shoulder pain	174	143	82.2	1.61	1.32

Leg pain	172	126	73.3	1.69	1.24
Neck pain	174	137	78.7	1.62	1.28
Muscle pain after exertion	171	140	81.9	1.87	1.53
Pain from brace	159	44	27.7	1.3	0.36
Eye irritation	172	115	66.9	1.3	0.87
Limited activity from pain	173	122	70.5	1.83	1.29
Cramping	173	113	65.3	1.26	0.82
Foot pain	174	90	51.7	1.33	0.69
Hip pain	174	110	63.2	1.97	1.25
Arm pain	173	99	57.2	1.54	0.88

Legend Table 5:

274 questions about symptoms in FSHD divided into 2 versions of the surveys. Number of Responses: Number of patients who answered the question. Number affected: Number of patients who reported to be affected by the symptom. Mean response: Mean impact score. Population impact score: product of the percentage of participants with the symptom and its average impact score