

Supplemental Table 1. Symptoms and Themes of Patients with CMT1A

Symptom	Total Number of Quotes	Total Number of Patients	Theme
PHYSICAL HEALTH			
Hand weakness	13	7	Hand and Finger Weakness
Decreased dexterity	9	6	Hand and Finger Weakness
Weak grasp	7	5	Hand and Finger Weakness
Dropping objects with hands	7	6	Hand and Finger Weakness
Difficulty manipulating small objects	4	5	Hand and Finger Weakness
Difficulty opening jars or bottles	3	2	Hand and Finger Weakness
Difficulty opening hands	2	2	Hand and Finger Weakness
Difficulty opening clasps on chains	2	1	Hand and Finger Weakness
Difficulty holding a pen	2	2	Hand and Finger Weakness
Decreased hand dexterity after being in cold weather	2	1	Hand and Finger Weakness
Hand tremor	1	1	Hand and Finger Weakness
Clumsy with hands	1	1	Hand and Finger Weakness
Increased hand weakness when tired	1	1	Hand and Finger Weakness
Trouble turning objects with hands	1	1	Hand and Finger Weakness
Subtotal for theme	55		
Decreased ability to carry a heavy load with arms	4	3	Proximal Upper Extremity Weakness
Arm weakness	1	1	Proximal Upper Extremity Weakness
Shoulder weakness	1	1	Proximal Upper Extremity Weakness
Subtotal for theme	6		
Requirement for ankle foot orthoses (AFO)	6	6	Foot Weakness
Foot drop	5	4	Foot Weakness
Ankle weakness	5	4	Foot Weakness
Foot weakness	4	4	Foot Weakness
The need for orthotic shoes	3	3	Foot Weakness
Required ankle surgery	1	1	Foot Weakness
Subtotal for theme	24		
Leg weakness	2	2	Leg Weakness
Difficulty getting up from the floor or ground	2	2	Leg Weakness
Difficulty getting up from lying down	2	1	Leg Weakness
Difficulty rising from a seated position	2	2	Leg Weakness
Subtotal for theme	8		
Impaired walking	26	13	Mobility and Ambulation
Falls	18	10	Mobility and Ambulation
Tripping	16	13	Mobility and Ambulation
Inability to run	11	10	Mobility and Ambulation
Difficulty with rough ground	10	9	Mobility and Ambulation
Difficulty walking long distances	10	6	Mobility and Ambulation

Difficulty going upstairs	10	6	Mobility and Ambulation
Decreased mobility	8	4	Mobility and Ambulation
Difficulty moving quickly	6	5	Mobility and Ambulation
The need for railing on stairs	6	5	Mobility and Ambulation
Difficulty going downstairs	6	4	Mobility and Ambulation
Need to be cautious walking	6	4	Mobility and Ambulation
Difficulty running	4	4	Mobility and Ambulation
Difficulty standing	4	3	Mobility and Ambulation
The need for assistive devices (cane, etc)	4	3	Mobility and Ambulation
Difficulty with ambulation	4	1	Mobility and Ambulation
Having to look at feet with walking	2	2	Mobility and Ambulation
Can't keep pace with friends	2	2	Mobility and Ambulation
Difficult walking in sand	2	2	Mobility and Ambulation
Fracture from falls	2	2	Mobility and Ambulation
Decreased coordination of legs	2	1	Mobility and Ambulation
Trouble with hills	2	1	Mobility and Ambulation
Loss of stability when walking	2	1	Mobility and Ambulation
Sense that walking is uneven	1	1	Mobility and Ambulation
Need to lift legs higher while walking	1	1	Mobility and Ambulation
Difficulty getting up quickly	1	1	Mobility and Ambulation
Trouble getting around when it is slippery	1	1	Mobility and Ambulation
Stubbing of toes	1	1	Mobility and Ambulation
Subtotal for theme	168		

MENTAL HEALTH

Cognitive Impairment	0	0	Cognitive Impairment
Subtotal for theme	0		
Fear of disease progression	15	9	Emotional Distress
Depression	8	8	Emotional Distress
Anxiety	6	6	Emotional Distress
Frustration	9	6	Emotional Distress
Fear of falling	7	6	Emotional Distress
Moodiness	3	3	Emotional Distress
Fear	2	2	Emotional Distress
Self pity	2	2	Emotional Distress
Awareness of disease progression	2	2	Emotional Distress
Fear of injury from sports	2	2	Emotional Distress
Anxiety about going places to do things	2	2	Emotional Distress
Damaged pride with loss of independence	1	1	Emotional Distress
Fear of stairs	1	1	Emotional Distress
Fear of escalators	1	1	Emotional Distress
Fear of an inability to perform job	1	1	Emotional Distress
Embarrassment	1	1	Emotional Distress
Anger	2	1	Emotional Distress
Fear of injury or death while being alone	2	1	Emotional Distress
Sadness	1	1	Emotional Distress
Hopelessness	0	0	Emotional Distress
Grief	0	0	Emotional Distress

Fear of being trapped in a burning building	1		Emotional Distress
Subtotal for theme	69		
Embarrassment of gait	5	3	Body Image
Embarrassment of braces	4	2	Body Image
Loss of hand muscle	4	3	Body Image
Clothing Restriction due to Braces	3	3	Body Image
Feeling different from other people	3	3	Body Image
Loss of leg muscle	3	3	Body Image
Weight gain because can't exercise	2	2	Body Image
Embarrassment of feet	1	1	Body Image
Loss of foot muscle	1	1	Body Image
Scoliosis	1	1	Body Image
Subtotal for theme	27		
SOCIAL HEALTH			
Lack of understanding of CMT by others	6	4	Social Role Limitations
Reliance on family members	6	4	Social Role Limitations
Impaired job performance	5	5	Social Role Limitations
Lack of job secondary to disability	4	4	Social Role Limitations
Unable to live in certain places	3	3	Social Role Limitations
Unable to go places	3	3	Social Role Limitations
Inability to do physical things other people can do	2	2	Social Role Limitations
Having to be cautious when going places	2	2	Social Role Limitations
Braces limit job opportunity	1	1	Social Role Limitations
Inability to go out	1	1	Social Role Limitations
Impaired sexual function	1	1	Social Role Limitations
Difficulty helping with family chores	1	1	Social Role Limitations
Unable to lift children	1	1	Social Role Limitations
Subtotal for theme	36		
Unable to participate in fun activities	7	7	Social Role Dissatisfaction
Dissatisfied with social interactions	6	4	Social Role Dissatisfaction
Social isolation	6	5	Social Role Dissatisfaction
Impaired interaction with family members	5	4	Social Role Dissatisfaction
Concern about being judged by others	3	2	Social Role Dissatisfaction
Feeling excluded	3	3	Social Role Dissatisfaction
Impaired interactions with friends	3	3	Social Role Dissatisfaction
Discrimination at work	2	2	Social Role Dissatisfaction
Difficulty with others who don't understand my limitations	2	1	Social Role Dissatisfaction
Difficulty getting around in large crowds	1	1	Social Role Dissatisfaction
Subtotal for theme	38		
CMT1A SPECIFIC ISSUES			
Inability to play sports	13	9	Specific Activity Impairment
Increased time to complete an activity	7	6	Specific Activity Impairment
Decreased ability to button	6	5	Specific Activity Impairment
Difficulty cooking	6	5	Specific Activity Impairment

Trouble taking care of the home	6	5	Specific Activity Impairment
Increased planning to complete an activity	6	3	Specific Activity Impairment
Difficulty cleaning the house	4	3	Specific Activity Impairment
Difficulty dancing	3	3	Specific Activity Impairment
Decreased ability to use zippers	3	3	Specific Activity Impairment
Difficulty using a ladder	3	2	Specific Activity Impairment
The inability to go certain places	3	3	Specific Activity Impairment
Trouble opening bottles	2	1	Specific Activity Impairment
Difficulty typing	2	2	Specific Activity Impairment
Difficulty swimming	2	2	Specific Activity Impairment
Impaired ability to exercise	2	1	Specific Activity Impairment
Difficulty vacuuming	2	2	Specific Activity Impairment
Difficulty painting a picture	2	2	Specific Activity Impairment
Difficulty cutting the lawn	2	2	Specific Activity Impairment
Difficulty tying shoes	2	2	Specific Activity Impairment
Can't walk in snow	2	2	Specific Activity Impairment
Difficulty biking	2	2	Specific Activity Impairment
Difficulty turning pages	1	1	Specific Activity Impairment
Decreased ability to use hand tools	1	1	Specific Activity Impairment
Trouble holding a piece of paper	1	1	Specific Activity Impairment
Difficulty using scissors	1	1	Specific Activity Impairment
Difficulty doing laundry	1	1	Specific Activity Impairment
Difficulty using a step ladder	1	1	Specific Activity Impairment
Difficulty shoveling	1	1	Specific Activity Impairment
Difficulty using a hammer	1	1	Specific Activity Impairment
Difficulty cutting objects	1	1	Specific Activity Impairment
Difficulty using utensils	1	1	Specific Activity Impairment
Difficulty washing dishes	1	1	Specific Activity Impairment
Difficulty hiking	1	1	Specific Activity Impairment
Difficulty operating a motor vehicle	1	1	Specific Activity Impairment
Difficulty preparing food	1	1	Specific Activity Impairment
Difficulty skiing	1	1	Specific Activity Impairment
Difficulty gardening	1	1	Specific Activity Impairment
Difficulty using a lighter	1	1	Specific Activity Impairment
Difficulty golfing	1	1	Specific Activity Impairment
Subtotal for theme	98		
Balance problems	19	12	Balance Impairment
Problems standing in the shower	3	3	Balance Impairment
Impaired coordination	2	2	Balance Impairment
Difficulty walking with objects	2	2	Balance Impairment
Subtotal for theme	26		
General fatigue	10	7	Fatigue
Fatigue when walking distances	4	2	Fatigue
Muscle aching with activity	1	1	Fatigue
The need for prolonged recovery time	1	1	Fatigue
Muscle fatigue	1	1	Fatigue
Subtotal for theme	17		

Cramps	7	7	Pain
Leg pain	4	3	Pain
Foot pain	4	3	Pain
Back pain	3	3	Pain
Pain when walking	3	2	Pain
Diffuse pain	2	2	Pain
Joint stiffness	2	2	Pain
Calf pain	2	2	Pain
Muscle stiffness	2	2	Pain
Burning pain with cold weather	2	1	Pain
Pain with orthotic use	1	1	Pain
Burning pain	1	1	Pain
Lack of flexibility	1	1	Pain
Pain in feet when standing up	1	1	Pain
Shoulder pain	1	1	Pain
Muscle spasms	1	1	Pain
Subtotal for theme	37		
Restless legs	3	2	Sleep Disturbance
Sleep apnea	1	1	Sleep Disturbance
Subtotal for theme	4		
Foot numbness	6	4	Numbness
Numb feet with cold weather	4	3	Numbness
Numbness	3	2	Numbness
Burns or injuries due to numbness	3	3	Numbness
Hand numbness	2	2	Numbness
Leg numbness	2	2	Numbness
Problems knowing where legs are	1	1	Numbness
Tingling	1	1	Numbness
Unable to feel temperature of water	1	1	Numbness
Cold feet	1	1	Numbness
Calf numbness	1	1	Numbness
Subtotal for theme	25		
Hearing loss	3	3	Central Sensory Impairment
Conversational hearing impairment	2	2	Central Sensory Impairment
Can't hear high pitches	1	1	Central Sensory Impairment
Difficulty with depth perception	1	1	Central Sensory Impairment
Subtotal for theme	7		
Heartburn	3	3	Gastrointestinal Issues
Constipation	3	3	Gastrointestinal Issues
Subtotal for theme	6		
Trouble swallowing liquids	2	2	Problems Swallowing or Choking
Trouble swallowing	1	1	Problems Swallowing or Choking

Subtotal for theme	3		
Regret over passing disease to children	3	3	Genetic Issues
Fear of passing disease onto children	2	2	Genetic Issues
Difficulty with childbirth	1	1	Genetic Issues
Subtotal for theme	6		
Total Quotes	660		
Total Number of Identified Symptoms	145		