

Don't Forget The Women

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<https://www.youtube.com/watch?v=V6fHTyVmYp4>

A Little About Me

- Physiotherapist
- Academic
- Interest in learning with use of narrative
- Importance of empathy
- Learning through listening



My Interest In KD/Carriers

- Physiotherapist with children with Duchenne Dystrophy and Spinal Muscular Atrophy
- Private practice people with long term conditions eg. Huntingtons Disease
- Belief that carriers show symptoms (now acknowledged for Duchenne Carriers)
- Chance meeting in a pub
- Film to teach about people with KD along side how to be involved in diagnosis and treatment of the gentlemen

Chris



What Did He Tell Me About Carriers?

My mother, I think she had symptoms. She would say my feet they are burning.

Why Did This Make Me Take Action?

- You are patients too
- Your physical, social and health care needs are not being met
- Carrier signs could help diagnosis in men
- You are carrying more than Kennedys Disease (burden of guilt, care, finance etc)

The Action

- Survey
- To follow interviews and film of ladies
- Aim publication

About this Survey

- Acknowledge findings from a small group of self selecting ladies
- Range of ages
- Different geographical locations
- YET so many similarities



Findings



1) Consent

- 38 ladies
- 100% Consent
- *Italic indicates direct quote* changed only if confidentiality/identification could be breached
- *Colours are used to separate comments not a code*
- *More ladies have completed*

2) At what age were you diagnosed as a carrier of Kennedy's Disease?

Age at Diagnosis 23 Respondents

- 7 15 15 17 18 22 27
- 30 30 32 32 35 36 36 37 38
- 54 55 59 60 62 65 65

Age Now 24 Respondents

- 22 28 28
- 30 32 33 36 37 38 38
- 40 40 41 44 48
- 54 55 56 60 65 67 68
- 71 79

3) How were you diagnosed?

25 Responses

- Assumption based on fathers positive diagnosis x 15
- Assumption based on male first cousin/brother/sons x 5
- Assumption based on carrier mum x 1
- Genetics/blood test x 9

4) Any comments relating to diagnosis?

14 Responses

Whatever minor symptoms I have may have nothing to do with KD

I knew of my diagnosis and my husband and I have chosen to move forward with IVF and embryo testing

I was officially tested to provide information to CooperGenomics, the company creating my DNA probe for embryo testing

4) Any comments relating to diagnosis?

14 Responses

I am very interested in sharing my symptoms

I began having symptoms in my late 30's, I am in my 60's and symptoms are getting much worse

By the time I had my official test done my oldest son had been diagnosed two years before. Two of my nephews 6 years before my son

4) Any comments relating to diagnosis?

14 Responses

My parents found out so much information by attending a conference in America. It was through this that I learnt I was a carrier. I then had issues dealing with being a carrier in the sense of when I'm ready to start a family and how this will actually affect me. Mum expressed concerns at Dad's yearly appointment at Nottingham Queens Medical, I was then given a genetic counselling appointment, as a family, as I was only 14/15. After this appointment there was no support

4) Any comments relating to diagnosis?

14 Responses

I had no referral or follow up or any professional input and I received a letter to state my blood results confirmed I was a carrier

When my son was diagnosed there was no information given by doctors for me other than “she must be the carrier”

I got the test in UK and live over seas, it may have been different if I lived in UK. In my letter it only states that I'm carrier and that I can pass it on. Nothing about me getting symptoms

4) Any comments relating to diagnosis?

14 Responses

Diagnosis was quick and decisive, there is a strong family history through my aunt and when my son developed symptoms I went for testing

I believe the patient's are left in the dark so in turn it leaves us carriers more in the dark

I'm still confused as to whether or not I should expect symptoms as a carrier as all health care professionals I've spoken to have said it doesn't affect females

4) Any comments relating to diagnosis?

14 Responses

Clinicians couldn't offer much help because they were unaware of KD before I saw them

Disappointed with neurologist who knew of it but really didn't offer much help. I had the feeling he was sceptical of my reported symptoms but was willing to hear my complaints and offer possible drugs. Disease is on my mother's side of the family which is very large. Extrapolating from positively diagnosed members, we have 11 members positive for KD

I have had no formal medical testing or medical advice

4) Any comments relating to diagnosis

14 Responses

When my father was diagnosed my parents did not tell me I was a carrier. I found out through researching the disease. I have never been formally diagnosed; however, my dad has had the genetic testing. I have never spoken to a health care professional about me as a carrier

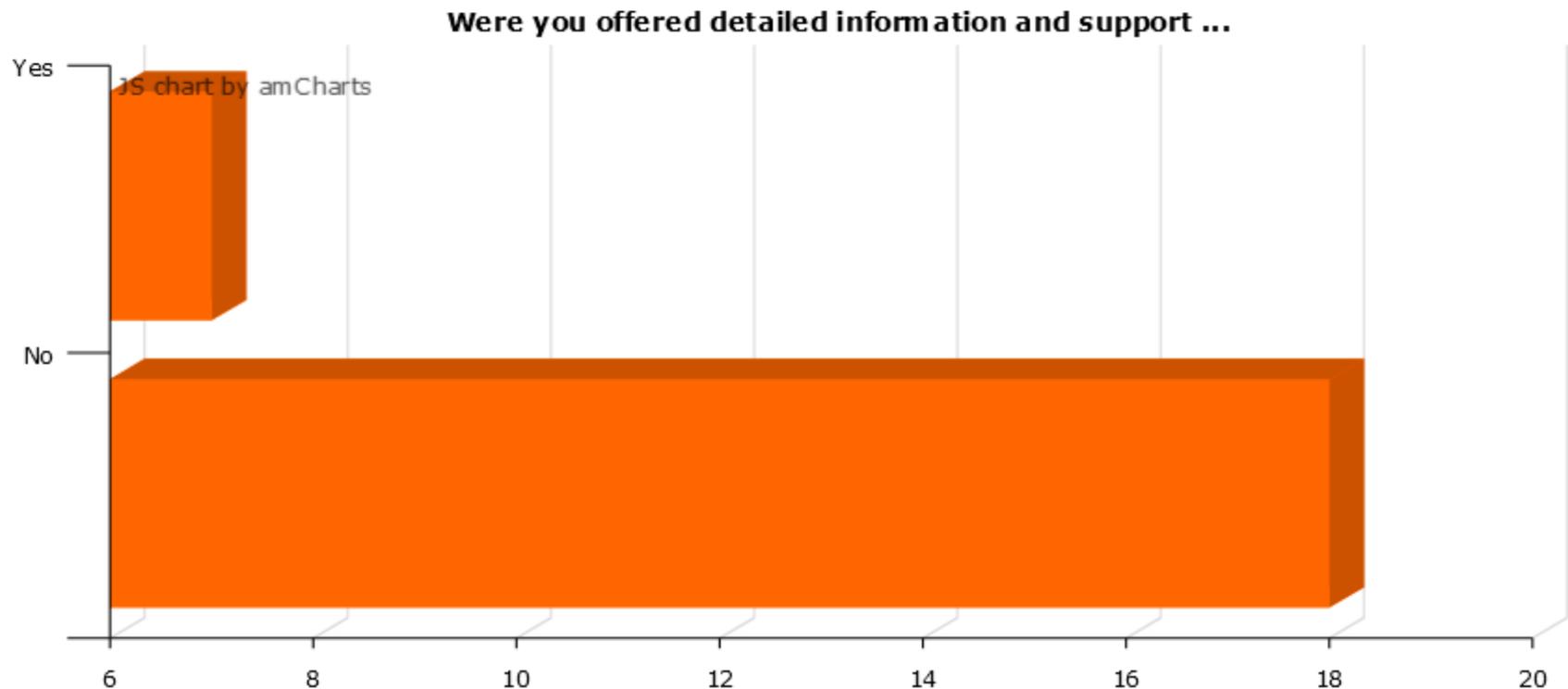
I was not officially diagnosed so I never had conversations with a medical professional

There doesn't seem to be much recognition for carriers

4) Were you offered detailed information and support?

25 Responses

Yes = 28% No = 72%



5) Please outline information and support offered
18 responses

None x 5 (and very little to my father x1)

Females are not supposed to have symptoms. I was advised to have daughters tested

*Appointments with a Genetics Nurse Specialist
Genetic counsellor session*

5) Please outline information and support offered
18 responses

I had done all of my own research. The test was proof for the NIH because I was asked to be in a study

Information came when I volunteered at NH Institute for a study

None of my doctors knew about KD. Even 3 neurologists have not been much help. I have sought out my own information

5) Please outline information and support offered
18 responses

I had genetic counselling through my Dads professor.

No referral was made for me to be seen

Support offered by Genetic Research Centre

5) Please outline information and support offered
18 responses

Father was told he had this via email with no support of his own, we had to figure it all out ourselves

Information about pregnancy tells me that women do not show illness

The genetic advisor never heard of the disease but offered support and testing/counselling for my daughter if she wanted it. She did get tested and was negative. This is my only child

5) Please outline information and support offered 18 responses

Counselling

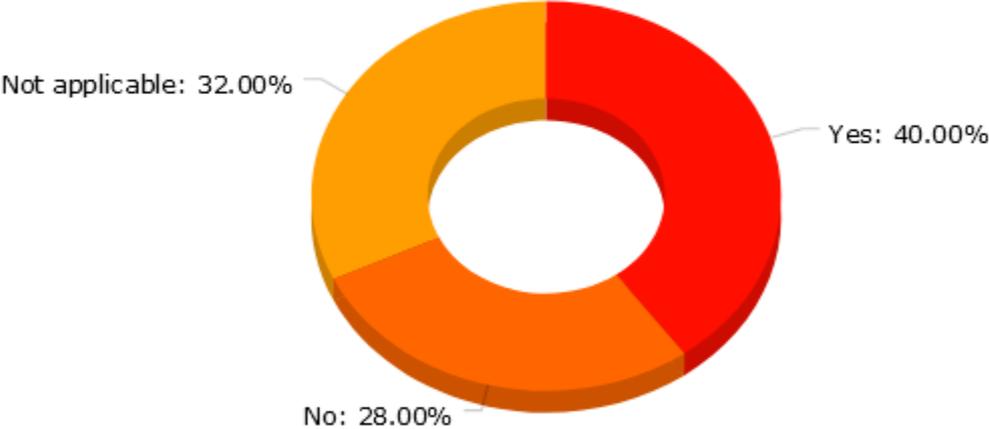
I live overseas and I got the blood test while back in UK visiting my dad. They advised genetic counselling when I got back home. As dad had just been diagnosed and didn't have many symptoms at the time the disease didn't seem that bad and I was only a carrier and told I wouldn't suffer I didn't feel the need

Drs affiliated with MDA diagnosed and offered support

6) Did those advising you already know about Kennedy's Disease?

JS chart by amCharts

Did those advising you already know about Kennedy'...



Yes

10

No

7

Not applicable

8

7) What were the clinicians' opinions about carrier symptoms?

The person doing my genetic blood test, unsure what his title was said "if you ever want a genetic disease this is the one to get, as it is mild". He said I wouldn't get any symptoms

I never knew anything about symptoms until my parents attended the annual conference for Kennedys

They said it doesn't affect carriers

7) What were the clinicians' opinions about carrier symptoms?

Said there could be some similarities to sons

Carriers weren't thought to have symptoms except in very rare cases

Slight symptoms but nothing like males

7) What were the clinicians' opinions about carrier symptoms?

It was not mentioned

They don't exist

Tells me that women do not show illness

Very open and interested

7) What were the clinicians' opinions about carrier symptoms?

Rare for carriers to have symptoms but not unheard of

Non existent

That they don't exist

Dads neurologist tells him that women don't have symptoms

7) What were the clinicians' opinions about carrier symptoms?

Father was told we would have no symptoms

That symptoms should be mild however given the severity of my symptoms he made a referral to a Neurologist for baseline assessment. However I remain on a waiting list now for over 2 years

7) What were the clinicians' opinions about carrier symptoms?

I have briefly asked in later years (I am 72 now) but physician has not seen reason to worry since I am female

That they wouldn't show (if ever) until much later in life and would be far less than that of my Father's symptoms

My prenatal genetic counsellor asked me questions about KD. Not familiar with it

7) What were the clinicians' opinions about carrier symptoms?

None

Carriers won't get symptoms

Most research said carriers had only slight symptoms so my primary doctors pretty much blew it off!

At first I was told carriers did not have symptoms I asked, who was listening? I am an RN and no one seemed interested

8) When you were diagnosed you were given clear explanations about whether you may have/develop symptoms

- 61% Strongly Disagreed
- 21% Disagreed
- 18% Agreed

You were given opportunity to have all
of your questions/concerns answered
by your Doctor//Nurse/other
Professional

- Strongly Disagree 41%
- Disagreed 27%
- Agreed 23%
- Strongly Agreed 9%

Overall, you were satisfied with the way you were cared for at diagnosis

- Strongly Disagreed 36%
- Disagreed 23%
- Agreed 32%
- Strongly Agreed 9%

Your wait time for the diagnosis was reasonable.

- Strongly Disagree 13%
- Disagree 20%
- Agree 43%
- Strongly Agree 24%

11) Do you have any signs or symptoms that you attribute to being related to being a Kennedys Disease carrier

Yes 86.9%

No 13.04%

12) Which of the following have you experienced?

Based on the symptoms experienced by the men

Symptom	% Daily	% Once weekly	% Once monthly	% Occasional	% Never
Fatigue	38	14	0	27	14
Muscle spasm	10	33	5	23	23
Pain	36	9	5	23	23
Tremor	6	6	0	27	54
Burning feet	14	10	0	27	54
Unusual shape tongue	15	0	0	0	86
Postural changes	10	5	0	5	68
Swallowing difficulties	40	32	9	4.5	9
Weakness	24	0	5	38	42
Cough	14	14	0	22	45
Aspiration of food	9	5	9	40	36

Symptom	% Daily	% Once weekly	% Once monthly	% Occasional	% Never
Back pain	27	13	0	27	15
Breathing problems	0	0	5	14	76
Pins and needles	19	9	6	50	18
Lack of balance/instability	28	14	5	36	28
Falls	0	0	5	14	81
Difficulty walking up hill	10	5	0	23	57
Difficulty walking on uneven ground	9	6	0	23	56
General difficulty with walking	9	5	0	22	52

Symptom	% Daily	% Once weekly	% Once monthly	% Occasional	% Never
Cramps	14	33	10	42	0
Facial muscle twitch	5	14	0	38	42
Arms and legs twitch	14	10	5	33	33
Body muscle twitch	14	10	0	23	52
Issues following anaesthesia	0	0	0	15	85
Muscle wasting	0	0	0	10	90
Hiatus hernia	0	0	0	5	95
Speech difficulties	0	0	5	32	78
Shaky movements when holding certain postures	9	5	5	28	47

Symptom	% Daily	% Once weekly	% Once monthly	% Occasional	% Never
Enlarged calf muscles	24	0	0	0	77
Localised numbness	5	5	10	25	55
Absent reflexes	5	0	0	0	95
Other	60	0	0	0	40

13) Outline any issues that you experience that you think could be related to being a carrier

Twitching of the face muscles, regular back ache, sudden body shake all over

Balance issues. Pins and needles. Slowed walking (i do exercise). Back pain

I get neck cramps/charley horses in my throat and neck

13) Outline any issues that you experience that you think could be related to being a carrier

I have developed muscle tremors and spasms especially at night. Muscles feel like they are constantly tense and will not relax. Because of this, I have difficulty falling asleep. I have hand tremors that affect my writing. If I am reading aloud, my mouth and tongue become tired and I start slurring my speech. I am constantly fatigued and feel like I could sleep for days

I have had swallowing difficulties since I was a child. I find I now choke a lot on food and have to bring it back up. I am getting pains in my arms and legs. I also have spells of vertigo

13) Outline any issues that you experience that you think could be related to being a carrier

Hip pain, unsteady gait. Sore throat, difficult swallowing

Muscle pain

Laryngospasms

I get regular muscle cramps in my jaw below my chin. I also get muscle spasms in my stomach on occasion

I get small leg cramps, but it could very well be due to many other causes

I have hypermobility where my joints dislocate on their own and most of the time I put them back in. My calves are huge. I sometimes gag on food. I can not walk unaided when I am out. I get stomach cramps a lot

Chest muscles very fatigued when singing in church

13) Outline any issues that you experience that you think could be related to being a carrier

I've constantly had aching legs but I'm not sure if this is due to Kennedy's or the fact that I am a bit unfit (is probably easy to attribute things to Kennedy's now I know I am a carrier!)

Chest muscles very fatigued when singing in church

Restless legs since childhood

Mainly cramps, tremor, twitching, fatigue. D contracture in both hands and soles of feet

14) Do you have any other condition?

Slightly elevated blood pressure. Heart palpitations

Frequent nausea

Fibromyalgia, which is under control. Moderate depression

I have high cholesterol. I suffer from reflux

I have Raynaud's

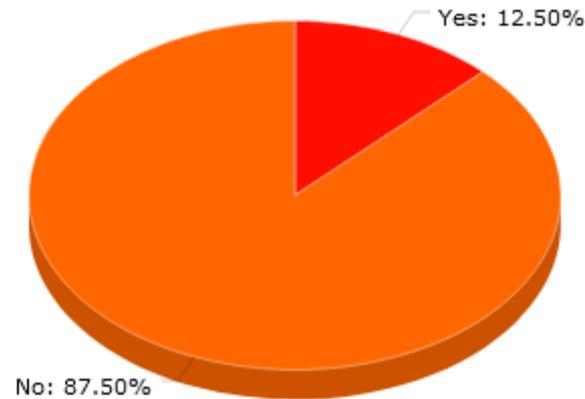
Osteoarthritis

I had large breasts and had reduction surgery 12 years ago

15 : Have you been offered any treatment from a health professional?

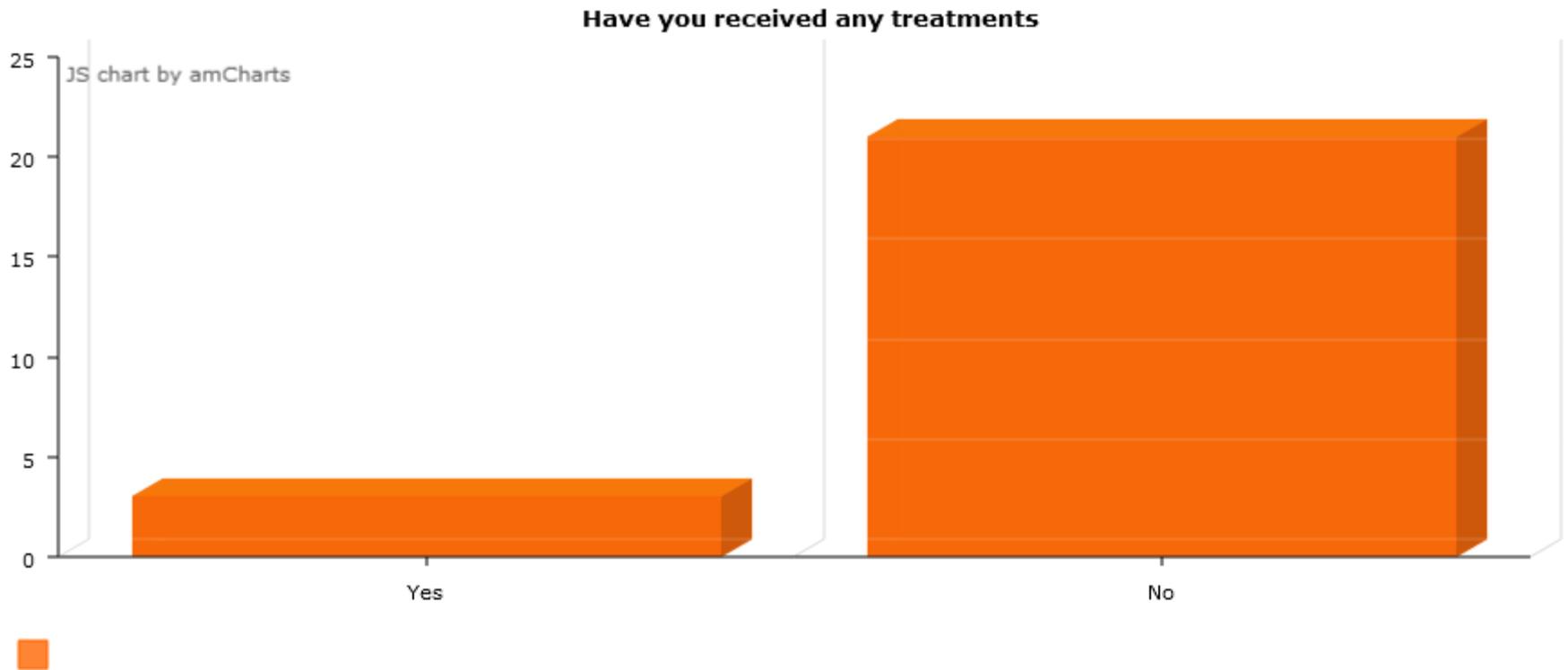
JS chart by amCharts

Have you been offered any treatment from a health ...

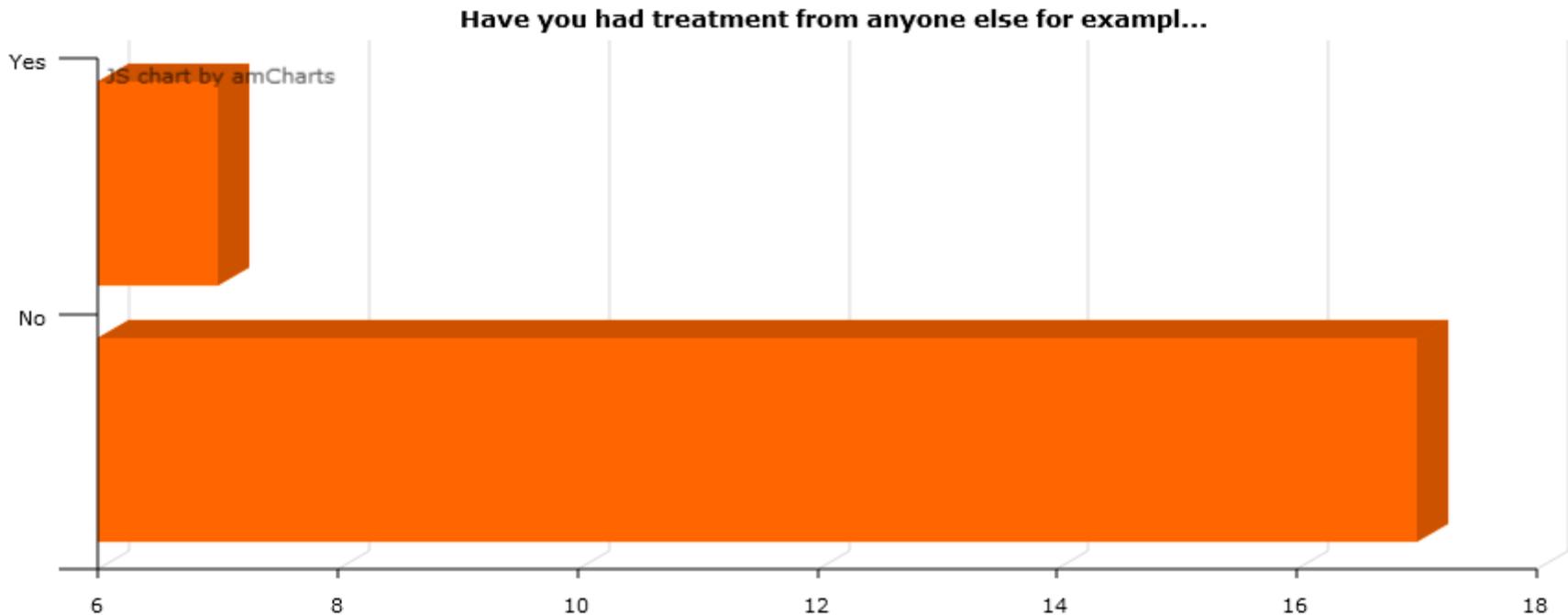


Yes 3 No 21

16 : Have you received any treatments



17) Have you had treatment from anyone else eg sports therapist, beautician, alternative practitioner?



18) Please offer brief details of the intervention

Chiropractor, no improvement. Have tried several meds for the neuropathy, unable to tolerate

I have acupuncture privately. My practitioner uses Chinese medicine acupuncture and I find this very effective. Previously had physiotherapy, I found the therapist unhelpful

Physical therapist for back and for balance

Massage

Physio and massage therapist... just massaging the muscles helps

I had acupuncture in my back and shoulders

19) Have you devised any treatments or strategies to help relieve your symptoms

Yes 18.18%

No 72.73%

20) If you have devised strategies please briefly outline these

Acupuncture Yoga Counselling

Rest

Light walking everyday, can't do cardio any more. Healthy eating, I find dairy, sugar and alcohol make my symptoms worse

I schedule a massage monthly. I try to exercise/swim in heated therapy pool. Sauna when available. Marijuana/CBC, ibuprofen, alleve Magnesium. I consult with my dr. Who is supportive of my strategies

Tylenol

21) Other comments

One of my daughters is a carrier. This is one of the factors in her decision not to have children The other one has not been tested but is not planning to have children anyway

I am greatly relieved that they are finally looking at the carriers and attending to them. I also look forward to the time when they can correctly diagnose the disease, and look at the causes. I have two sons who have it and potentially 4 great grandsons who could be diagnosed

Depression is a big issue, guilt related for passing the gene on to my two sons , my daughter has not been tested.

I have had swallowing difficulties my whole life, that could never be explained. My doctor told my mum it was in my head. I've only really started to get what I think are Kennedy related symptoms in the last 5 years. My dad was on life support twice last year and is now peg fed, but doing really well. He suffers a lot, but doesn't think about me as a carrier and doesn't really believe my symptoms are related. I'm the lucky one apparently! I haven't seen a specialist yet as medical is so expensive and effects your insurance with any diagnosis. If I still lived in UK I would probably access better support and care

21) Other Comments

The more info the better with all aspects of this disease

While I have not ever been officially diagnosed as a carrier, I feel that some issues I experience are related and do plan to be tested at some point since my brother is deeply affected

I wonder how the other carriers solve their pregnancies, whether they are going to artificial insemination

Very frustrating to read the medical literature that discounts carrier symptoms

21) Other Comments

I haven't been tested. My dad's symptoms mimic ALS much closer than Kennedy's disease

When I said to the doctor I could have symptoms of KD he said i don't think so he didn't know any thing about it!! It would be nice to be recognised as some one that might be showing symptoms of KD

Notice symptoms worsen for me at rest. Muscle twitching and cramping especially if on my feet most of day or dancing etc

I feel like a completely different person in the last couple of years. I have always been active, strong and an early riser. Now I feel weak, tired and run down. My muscles cramp, twitch and ache all the time. I shake and my speech is starting to slur when teaching. Most doctors are not aware of KD and therefore it makes it difficult to find relief

What Next

- Individual interviews
- Film of ladies
- Can anyone sing?
- Article publication
- Keep pushing on



Thank You

To everyone who participated a big thank you
For listening now a big thank you

Remember you are woman you are strong! If women of the world Unite
we can make changes!



Questions?

Jill Wickham

email me at
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- Physiotherapist
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