Coping with a neuromuscular illness, with a particular focus on Facioscapulohumeral muscular dystrophy (FSH)

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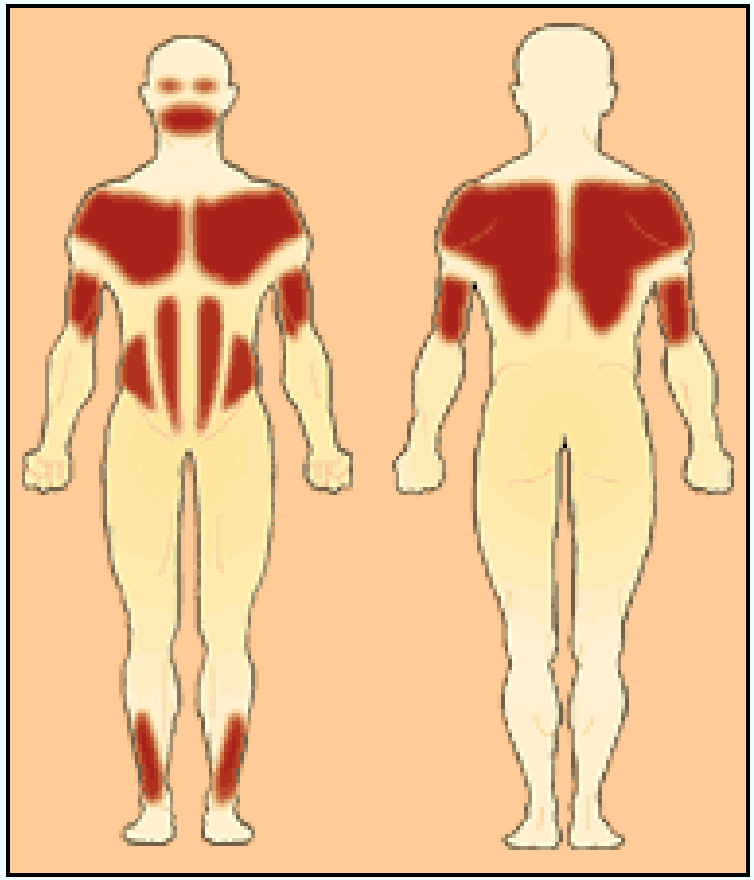
♦ Facioscapulohumeral muscular dystrophy:

- Major symptom: the progressive weakening and loss of skeletal muscles in the face (facio), shoulder area (scapulo) and upper arms (humeral). Early weakness of the muscles of the eye & mouth (smile, pucker, whistle), with weakness in the shoulder muscles, are distinctive of FSH. Other skeletal muscles may also eventually weaken - the muscles of the foot, hip, and abdomen. It does not shorten one's lifespan. Heart problems are rare. It doesn't affect the brain.
- The symptoms & progression of FSH are highly variable. Symptoms generally appear by age 20 and progress slowly.
- Most people show asymmetrical weakness (1 side weaker).
- About 20% of persons with FSH will require a wheelchair.
- FSH occurs in about 5 per 100,000 people.
- In Canada, there are 340 people with FSH registered with MDC (about 4.3% of MDC clients). This makes FSH the 5th most common dystrophy.

♦ Facioscapulohumeral muscular dystrophy:



♦ FSH is a genetic disease:

- FSH is a genetic disease caused by a mutation in the DNA of chromosome # 4 (a deletion – some DNA is missing).
- The missing DNA isn't part of a gene, but it still somehow causes FSH (perhaps it controls some muscle genes). Very complex problem - little is known about how this DNA works.
- FSH is an autosomal dominant disease 70% to 90% of the time – this means a son or daughter of an affected person is at a 50% risk of inheriting the defect.
- In about 10% to 30% of cases, the mutation is sporadic (the mutation was not passed on, it arises as a new mutation).
- The size of the deletion is thought to be related to the severity of the symptoms (but the severity of FSH in a parent does not predict the severity in an affected child).
- There is a genetic test for this deletion (& a prenatal test).

♦ Introduction:

- In this presentation I will discuss some of the impacts of neuromuscular illness (NMI) and focus on various aspects of coping.
- There are many general aspects to NMI, however, each specific type of illness also presents its own unique features & challenges.
- The impact of NMI is also unique for each of us:
  - The same muscular illness may affect different people in different ways. Many will start at different times and show different rates of progression.
  - People <u>experience</u> their illness in a unique way.
  - People <u>react</u> to their illness in a unique way.
  - People learn how to <u>cope</u> in a unique way.

- NMIs are complex and chronic disorders:
  - Currently, most NMIs lack effective treatments.
- Our reaction & response has a major impact on how we will live and cope with our illness.
- Many factors determine the impact of a NMI:
  - What are the symptoms? When did symptoms emerge? How does it affect you physically? How does it impact your lifestyle? Affect mobility? Balance? Basic functions? Does it progress? How fast? Is it treatable? What impact does treatment have? Do you have a strong support system?

Three Types of Coping Are Important:

- 1). Personal Coping - A Private Issue:
  - We must face up to the fact that we have a chronic illness & that our lifestyle will be affected.
  - This is largely a personal and private process.
  - How we come to see our illness is critical to our subsequent attitude and our attitude is critical to how we will react and cope in the long term.
    - This is the most important point discussed here.

♦ 2). Coping With The Medical Aspects:

- Our medical team is an ongoing aspect of our dealing with our illness, both in diagnosis & after.
- The ongoing care, compassion & advice of our medical team is vital, even in “untreatable” conditions.
- Doctors and other health care providers:
  - “Know” about these illnesses academically but don’t have the “benefit” of experiencing them.
  - Their tools: science & empathy.
  - They must provide us with a context for our illness.
  - Feeling that “our medical team” understands and is available when needed is an important support for us.

♦ 3). Social Coping - Dealing With Others:

- Our family & friends will often have a hard time dealing with our illness.
- We often must educate others about our illness.
- A common reaction: “But you don’t LOOK sick.”
- Overly “helpful” friends may be a problem.
- Sometimes friends can’t deal with our illness and leave us.

- Psychological Stages of Coping with Illness:
  - Some authors describe various psychological stages & give charts of different stages we will face.
  - In my opinion, there is no one pattern of stages that fits everyone, look for your own pattern of reactions.
- Most people do share three basic phases:
  - 1). Before going to the Doctor:
    - We have symptoms but don’t know what’s wrong.
  - 2). The diagnostic phase:
    - Often a long, frustrating and difficult process.
    - May or may not conclude in a clear diagnosis.
  - 3). After the diagnostic phase:
    - Long-term management and adaptation.

♦ What Could That Be From?

– As our symptoms develop, we see that something is wrong with us (or with our child).

– Common reactions during this initial period:

- Uncertainty: symptoms create feelings of fear & anxiety.
- Denial: ignore problems & think nothing is wrong.
- Catastrophize: imagine the WORST thing that it could be.
- Feel guilt: “I must have done something to cause this.”
- Superstition: “If I just do this, I’ll get better.”
- Blame: look for someone or something to blame.
- Crisis: feelings can build and erupt into crisis.

- In adult patients, the early phases of a muscle disease may unfold slowly and we often go back and forth in our imagination from: “I’m O.K.” to “Oh my God, something really is wrong.”
- This is often a period of immobility: we feel “stuck” and many people don’t seek help immediately.
- “Sometimes it is better not to know” versus “Whatever it is, I have to face it.”
- It takes a lot of courage to end this phase by going to see a Doctor and starting the diagnostic process.

- Many muscle patients report having to fight to be heard & for the Doctor to take them seriously.

- NMIs are known for being hard to diagnose:
  - Many are initially undiagnosed or misdiagnosed.
  - Diagnosis is usually a long, difficult, complex & slow process with many steps - we must be patient.
- ***"***I'm not ***crazy*** . . . and I'm not just ***lazy:"***
  - Many people are initially "put off" by Doctors as "imagining things" or as simply being lazy.
  - We know our symptoms & we know when things are wrong, no matter how strange our story sounds.
- We have a serious & chronic (life-long) illness:
  - We must now play a new role: as long-term patients.
  - Just as our medical team are professionals, we must become **professional patients.**

- The diagnosis is important as it forecasts our expectations & determines future management:
  - We must feel that our diagnosis is correct:
    - What evidence was used to make your diagnosis?
    - Always seek a second opinion of the evidence.
- Often a NM diagnosis has many implications:
  - People need time for the diagnosis to “sink in.”
  - Many people will want to learn about their illness.
  - If necessary, ask to see the Doctor again if questions come up.
  - Ask questions until YOU feel that you understand whatever YOU need to understand.

- Diagnosis is a mixture of evidence, the Doctor's experience, and his or her attitudes & intuition.
- Many people run into "roadblocks" in diagnosis:
  - Don't take problems personally & try not to get upset or frustrated: it simply doesn't help things.
  - Consistently return to the questions, evidence & issues that are important in making your particular diagnosis until YOU feel satisfied.
- Different Doctors may have different ideas about what's wrong and what to do about it:
  - Different *types* of Doctors may also have different opinions based on their training.

- The “final” diagnosis may remain uncertain:
  - Two Doctors may make a different diagnosis or suggest different treatments, often with the same degree of confidence.
  - It’s up to the patient to decide what to do about conflicting recommendations – a difficult situation.
- NMIs are not well understood & few effective treatments are available: a depressing reality that both Doctors and patients have to face:
  - “No treatment” does not mean no help - we need advice & support from our medical team.
  - Both the patient & the Doctor have to deal with these issues and both need to develop an attitude of “compassionate & supportive realism.”

- The “final” diagnosis of FSH:
  - *Sometimes* FSH is diagnosed as other NMIs.
  - FSH usually displays a fairly clear & unique pattern of muscle weakness (it’s easier to diagnose).
  - Genetic testing can confirm ~95% of FSH cases.

- There is no “treatment” for FSH:
  - Management is focused on dealing with symptoms and learning to cope with specific weakness.
  - Braces are sometimes used for the ankles and occasionally surgery is used to help with the shoulder area, to give more movement to the arms.

- Ongoing Medical Support:
  - Ideally, patients with NMIs will have an ongoing & open relationship with their medical team after the initial diagnostic phase ends:
    - As we age & our NMI progresses, things change.
  - We need to keep learning and keep monitoring our symptoms as they unfold with the following in mind:
    - Was the initial diagnosis correct?
    - Emerging complications: detected & dealt with?
    - New tests may come out, do they apply to me?
    - Ongoing implications for physical rehabilitation?
    - Practical occupational and home care advice?

- Self-education:
  - We need to “let the Doctor be the expert,” however, a major trend in medicine is **patient involvement** - we need to understand enough to help us make **informed decisions** about our care, based on the **evidence** (another major trend in medicine).
  - Many people believe that patients should learn all they can about their particular illness.
  - Staying current with medical research and information often helps people feel hope: “something is being done, even if it might not help me now.”

♦ Information and Support:

- Learn where to get information on your illness:
  - Internet? Associations? MDC? Library? Other sources?
- Seek out associations and support groups:
  - Even with rare disorders, there are many others like you.
  - Talk to people about their experiences & problems.
  - If other people can find THEIR ways to cope, YOU and I can also find OUR own ways to cope.
  - Other people are a great source of information, practical advice & emotional support.
  - Don’t be afraid to talk to people & to ask questions.

- Advocacy: “Taking action in support of the patient & ensuring their needs are addressed.”
  - External advocates: Various associations (like MDC), your Doctor, other groups & friends can give us support, help us understand medical information & help us navigate through the medical system.
  - Self advocacy: We must also become our own advocates & be informed, firm & consistent:
    - We must be strong, stay positive & “look out” for ourselves and our needs.

♦ Illness “Covers” Normality:

– NMIs usually affect specific aspects of function:

- Our normal, unaffected functions continue on.
- Our symptoms often get 98% of our attention.

– It is important not to ignore our normal functions & our general health.

– NMIs may complicate the normal stages of growth:

- NMIs in children complicate growing up & make transitions (adolescence) difficult - parents need to be sensitive.
- In adults, NMI symptoms are imposed “on top of” the normal decline of muscle as we pass mid-life into old age.

♦ Emotions:

– “Is it normal to feel this way?”

- No matter how you feel, it is likely a normal reaction for you. There are no “right” or “wrong” feelings.

– To experience all of our feelings is a natural and important part of our lives and learning:

- It is important to try to be open to all of our feelings and to try not to deny them – depression is a NORMAL reaction.

- Depression:
  - The Roller Coaster: We will all have a wide range of feelings, some positive, some negative.
  - Depression is a major factor in persons living with a chronic illness.
  - If we find ourselves “too low” or low for “too long”, we may need to seek help to deal with things.
  - Fatigue (feeling tired) and stress are common in muscle disease and both add to our risk of depression.

♦ We Must Get Through And Rise Above:

- Stress, fear, anxiety & depression are NORMAL reactions to major life hardship & change.
- It's O.K. to be sad & feel sorry for our losses:
  - Let yourself "get through" these sad feelings.
- We need to actively balance the positive & the negative & not let the negative "take over us."
  - Try not to dwell on the sad & negative aspects:
    - Experience the sad feelings and then move on.
    - Actively add positive activities wherever possible.
- Try to learn from negative feelings:
  - When sad, what's a positive view or action you can come away with to better deal with life?

♦ What Will Tomorrow Bring?

- Often, people will imagine what <u>might</u> happen in the future & they develop strong anxiety.
- People with NMIs may get stuck on “future anxiety.”
- Many people deal with future anxiety using denial.
- Controlling future anxiety is the hardest point discussed here, but it is critically important:
  - We can not afford to be lost in denial or let anxiety over “what might happen tomorrow” ruin today.
- We need to focus on today & live for today:
  - We need to learn to relax, deal with what life gives us, learn new ways to adapt and “take life one day at a time.”

♦ Resources:

- Many people “sell themselves short.”
  - We often have more resources than we think.
- The most important resources we have are:
  - Our self-image and ourselves.
  - Our intelligence, common-sense and personality.
- I am still me, (albeit, me with this illness):
  - I haven't lost myself - I have not become my illness.
- Our self-image & resources are largely determined by our attitude: how we see life.
- Various Muscular Dystrophy groups are a major resource for support & information:
  - In Canada, MDC: http://www.muscle.ca/
  - In the USA, MDA: http://www.mdausa.org/

- Abilities:
  - Who is “able?” Who is “disabled?” Who decides?
    - 1 How do others see us? (we can’t control this).
    - 2 How do we see ourselves?(we can control this).
  - Many people are very different but still very able.
  - Don’t jump into the “disabled category.”
  - With NMI, abilities will change, however, with the right attitude, we can still be very able individuals.
  - As abilities change, so must our priorities:
    - My number one priority today is preventing falling and injuring myself.

- Accept and Adapt:
  - No one asks to be ill, it's not something we choose.
  - "It is what it is."
  - How we "see" this & deal with it makes a big difference to the kind of life we will live.
  - Coping with a NMI challenges us to face & to rise above many everyday problems and frustrations.
  - Accept that life will be different & continually adapt to get the most out of life as it unfolds.
- Human beings are wonderful at adapting:
  - Adapting: finding ways to "learn to live with it."
    - We need to see what is under our control in life.
    - We need to adapt to what we can't change.

- Fatigue:
  - Chronic muscle illnesses are very tiring and we need to plan our activities accordingly.
  - Even everyday, routine activities can be very tiring.
  - Anticipate when you will “run out of gas.”
  - We have to learn to work with the energy we have & learn not to overdo it – a VERY common problem.

- As some doors close, others open:
  - As our physical abilities change, we will need to curtail some of our activities:
    - For each activity you have to stop, look for a new one to do.
    - Our lives will change more than most people, so we need to focus more than most people on coping with changes.

♦ Attitude:

Definition: “A mental state involving beliefs and values leading to feelings and dispositions to act in certain ways.”

– People can influence and alter their attitudes by changing how they think about life.

– We need to see how the choices we make, concerning how we see life and how we live life, can make a huge difference, even when we have a chronic illness.

– Let’s be part of the solution, not part of the problem.

– Let’s focus on grace and compassion, not sadness and grief.

♦ NMI calls on us to be excellent managers:

- 1. Manage activities: Everything we do takes longer, is a bit more difficult and uses more energy.
  - Great advance planning is our best defense.
  - We need to increase efficiency, reduce redundancy (only do it once) & generally, be very well organized in life:
    - Activities in the house =versus= activities when going out.
    - What we can do ourselves =versus= when we need help.
    - Time management =versus= activity management.
- 2. Manage health: People with a NMI need to be very aware of their overall health - their health quotient.
  - Diet (amount & type of food), hydration, exercise.
  - Rethink nicotine, alcohol usage & all types of drug usage.
  - We need to encourage practices positive for health and minimize factors harmful to health.

- 3. Manage stress: Whatever causes it, stress creates a complicated series of mental & physical changes.
  - We need to be aware of our stress levels & learn how to manage stress as effectively as possible.
  - Uncontrolled stress may aggravate some NMIs.
- 4. Manage energy: Chronic muscle illnesses reduce our energy and we use more energy to live life.
- 5. Manage emotions: We need to be aware of our emotions and to try to “keep an even keel.”
- 6. Manage change: Accepting & coping with change is an ongoing part of life, especially as we get older.
  - We cannot avoid change, again, we have to have a positive attitude towards the changes in our lives.

- Good life management often involves many lifestyle changes.
  - Many of these are major & hard choices to make:
  - But remember, our goal is a vital one: to maximize our quality of life.
    - For example, we may HAVE to give up smoking or drinking to achieve a better long-term quality of life.
- All of these points equally apply to the parents and partners of persons living with a NMI.
  - Often the brunt of a muscle illness is “shared” by others in the family and they also need to be considered in terms of their life management.

♦ Biography of the Author:

- Bill Tillier:
- I developed a progressive muscular dystrophy in my forties.
- I was a forensic psychologist for 20 years. I am now on long term disability and volunteer my time for Muscular Dystrophy Canada and the Canadian Paraplegic Association. I have a very supportive partner, a supportive cat and many great friends.
- I have built a web page on muscle disorders with easy to read information: http://ibmmyositis.com/
- If you have any questions or comments, please e-mail me at: btillier@shaw.ca