DM Care and the Advocate's Role

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Goals of the presentation

- Understand why you need to take charge of your own health
- Get to know better your disease
- Tools for you and your healthcare professionals
Self-advocacy

- Understanding your strengths and needs, identifying your personal goals, knowing your legal rights and responsibilities and communicating these with others

  (DMC, 2010)

- Several contexts
Steps toward self-advocacy

• There are three important pieces to becoming an effective self advocate:
  • 1. know your needs
  • 2. know how to get what you need
  • 3. get what you need
Most patients with myotonic dystrophy are badly managed or, to be more precise, not managed at all.

Hilton-Jones, 1997

The current clinical services is probably inadequate and there are areas of the country that are poorly served.

Hill & Phillips, 2006

Why?
Multisystemic disease
Vision: Cataracts, retinal damage

Bone: Anomalies

Immune: Hypogammaglobulinemia

Skin: Pilomatrixomas

Respiratory System: Breathing difficulties, aspiration, sleep apnea

Endocrine System: Diabetes, low thyroid hormone levels

Reproductive System: Low testosterone levels, testicular failure and gonadal atrophy in men. Weakened uterine muscle, pregnancy-related complications, and gynecological problems in women.

Cognitive Function: Intellectual impairment, behavioral and psychological disorders, excessive daytime sleepiness

Cardiovascular System: Heart condition abnormalities, arrhythmias, cardiomyopathy

Gastrointestinal Tract: Swallowing issues, abdominal pain, irritable bowel syndrome, constipation/diarrhea, poor nutrition and weight loss, chronic infections

Muscle: Weakness, wasting (atrophy), myotonia, pain

MDF, 2012
Typical patients in GP office have generally two concerns to be addressed during their medical visit

(Flocke, SA et al, 2001)
Systemic involvement in DM1

- Muscular system
- Central nervous system
- Cardio-vascular system
- Visual system
- Respiratory system
- Gastro-intestinal system
- Genito-urinary system
- Reproductive system
- Metabolic & endocrine system

Need all to be reviewed during your follow-up visit

= 54 concerns
Different clinical presentations
Clinical presentation

- We can see patients with very few symptoms even if old.
- We can observe very young children severely affected by the disease.
- We can observe patients who fall more than once a week during their thirties.

All the same disease but not the same management.
Phenotypes in DM1

• A phenotype is any observable characteristic or trait.

• In DM1
  • Congenital
  • Childhood
  • Adult
  • Late or mild

• Large variation between patients even in the same family
Several places to receive services

- Family doctor office
- Neurologist clinic
- Neuromuscular clinic
- Rehabilitation centre
- Home services
- Community services
- Medical Home
Healthcare professionals

Do they know DM1?
Knowledge among healthcare providers

Initial training
- Nurses: no reference in textbooks
- Occupational therapy: few references with less than 1 paragraph in textbooks
- General practitioner: few hours on neuromuscular disorders

Actual practice
- Very few healthcare professionals will encounter a person with DM1 in their life
- Only one medical guideline in 1980 is published
Professional resources

• Excellent textbooks on DM1
  • Difficult to get access to it
  • GP needs to keep up to date about so many common diseases that very few time is available for DM1 if any
  • Same for most professionals

• Websites
  • Good general description but not enough to know what to assess, when, how and what to do with it.
You and your family is often the ones who know most
DM1 a muscle disease only?

Myotonic dystrophy is perhaps the most variable of all human disorders

(Sir Peter Harper, 2001)

Myotonic dystrophy is perhaps one of the most complex disorder to manage
DM1 is not a muscle disease only....
It leads to a clear socioeconomic disadvantage where almost every aspects of life are to some points disrupted
Participation in SLSJ, Québec, Canada

- 55% did not complete a high school degree
- Around 20% are currently working
- 42% are living below poverty line
- 63.5% have severe restrictions in doing major household tasks
- 26% have severe restrictions related to leisure activity
Environment

- Often reduced social support
  - Affected family members
  - Fewer friends
- Lack of targeted healthcare initiatives
  - Rare disease
- Poor access to healthcare services
  - Education
  - Money
  - Personality
Organisation of care

Different models of care which make the concept of standard of care difficult to achieve
Several professionals

- General practitioner
- Neurologist
- Cardiologist
- Pneumologist
- Nurse
- Nurse case manager
- Physical Therapist
- Occupational Therapist
- Social worker
- Nutritionist
- And so on...
How to improve your health follow-up?

- Becoming a family who is
  - Informed
  - Proactive
  - Involve in research
Getting informed : A start

What you should know about your health follow-up
Resources to improve your knowledge of your disease

- Management recommendations
  (Neuromuscular Disorders, 2010)
- Health checklist on MDF website
  (Neuromuscular Disorders, 2010)
- Integrated care pathway for your nurse
- Myotonic Dystrophy Foundation website
- The Facts (Sir Peter Harper)
Vision: Cataracts, retinal damage
Bone: Anomalies
Immune: Hypogammaglobulinemia
Skin: Pilomatrixomas
Respiratory System: Breathing difficulties, aspiration, sleep apnea
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Central Nervous System: main points

- Awareness of possible cognitive impairments
  - Understanding doctor’s recommendations
  - Developing strategies to put recommendations into place
- Depression and anxiety can be present and should be assessed
- Hypersomnolence (I sleep all the time)
- Fatigue
Strategies
Fatigue has been described as a major factor explaining difficulty in performing activities related to independent living, walking, working and leisure (Gagnon et al. 2008)
Fatigue

- Discuss with your doctor the difference between hypersomnolence and fatigue
- Potential medication
- Energy conservation technique with your occupational therapist
**Visual system**

- Ptosis
  - Drooping upper eyelid
  - Operation possible but with restriction
Cataracts affect vision which in turn can affect chances of falling, difficulty in driving and so on...
Respiratory System: Main Points

- Pneumonia
  - Most frequent cause of death
  - Influenza vaccine
  - Pneumococcal vaccine
- Sleep apnea and related treatment
- Anesthetic risks: make sure you inform your doctor
  - MDF website
  - Care card
  - Alert Bracelet
30% are smokers although pulmonary problems are the leading cause of death.
Solutions to stop smoking should be discussed with your doctor.
Cardiovascular system: main points

• Frequent cause of death

• Recommendations will vary among specialists but two main points
  • Need an annual ECG even if mildly affected done by the same clinic to look at progression
  • Follow-up in cardiology usually necessary at some point
Muscular system

Mathieu, J., et al., 2001; Mathieu, J., et al., 1992
Muscular system: main points

• Myotonia
• Walking limitations and related accessories
• Fall assessment and environmental related factors
• Transfers difficulty especially in the bathroom
• Exercise ????
Muscular system

- Myotonia
- Walking limitations and related accessories
- Fall assessment and environmental related factors
- Transfers difficulty especially in the bathroom
- Exercise
  - Physiotherapy presentation
Gastro-intestinal symptoms

- Malnutrition
  - 55% eat too much fat
- Silent dysphagia is often present which could lead to pneumonia
- Abdominal pain, constipation and diarrhea
- Fecal incontinence
- Treatment available with limited evidence
  - Presentation during the meeting
Genito-Urinary system

- Urinary incontinence
- Erectile dysfunction
  - Between 24.1% and 36.7%
  - Treatment is possible but caution is needed
- Obstetric risks
  - Need close follow-up as risks are present
- Gynecological problem
  - Painful menstruation
  - Irregular menstruation
Endocrine and metabolic system

- Obesity in DM1
  - 33.5% obesity
  - 27% overweight

- Blood tests
Why obesity management is even more important in DM1
Genetic aspects

- Transmission
- Anticipation
- Importance of genetic counselling
Transmission: Dominant disease

www.brusselsgenetics.be/p_453.htm
Anticipation

As myotonic dystrophy is passed from one generation to the next, the disorder generally begins earlier in life and signs and symptoms become more severe.
Genetic aspects

- Genetic counselling
  - Family planning
  - Risk for family members
  - Information from many website including UK Myotonic Dystrophy Support Group
  - Review information with your genetic counsellor or your doctor
- Insurance issues in USA
Once main symptoms are dealt with what’s left?

The role of the rehabilitation team
When do I need to see a rehabilitation professionals

When to see a physioterapist

Orthopedics & Physical Therapy
When to see an occupational therapist

Occupational Therapy
"We're not sure yet, but we think he may have been asleep at the wheel."
Driving and cataracts
Some recommendations

- Walking limitations
- Pain
- Educational support during school’s years
- Vocational rehabilitation services
- Budget services including fiscal information
- Home maintenance
- Social support
- Driving issue
- Participation in adapted leisure activities
- End of life issues
- Chronic Disease Management
- Neuromuscular Nursing Case Manager
Be proactive: A start

When you go to your GP brings your checklist
Keep a journal of your main symptoms
Bring a family member with you
Consult recognized websites
More tricks

- Create a filing system, containing the records you may need (e.g. medical, educational records) for easy access and reference.
- Prepare a checklist of thoughts, questions and concerns regarding the issue or problem you are advocating for. You can also use a computer, camera or audio recorder to keep your records.
And more.....

• Keep a journal that includes a log of all phone calls and meetings including the date and time, who you spoke with and what was said.

• Keep copies of all letters, emails, policies, procedures if they are relevant to your issue or problem. Keep a copy of all letters and emails you send to people and keep all original letters that you receive
Active in research

Be part of research initiatives when appropriate, patients’ registries, go to conferences, read association’s journals or newsletters.
References


References

• Myotonic Dystrophy : The Facts by Sir Peter Harper
• MDF website