# -USER GUIDE-

# INTEGRATED CARE PATHWAY TOOL FOR THE MYOTONIC DYSTROPHY TYPE 1 (DM1-ICP)

Maud-Christine Chouinard, RN, PhD[1,2,4]
Jean Mathieu, MD, M.Sc., FRCP (c) [1,3,4]
Mélissa Lavoie, RN, PhD (c).[1,4]
Nadine Leclerc, RN, BSc[1]
Aline Larouche, RN, MSc (c)[1]
Nancy Bouchard, RN, BSc[1]
Stéphane Jean, Neuropsychologist, MA[4]
Cynthia Gagnon, OT, PhD[1,3,4]

[1]Neuromuscular Clinic, Centre de réadaptation en déficience physique de Jonquière, Saguenay, Québec, Canada
[2]Health Department, Université du Québec à Chicoutimi, Québec, Canada
[3]Faculty of Medicine and Health Sciences, Université de Sherbrooke, Québec, Canada
[4]Centre hospitalier universitaire de Chicoutimi, Chicoutimi, Québec, Canada

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## DEVELOPMENT OF DM1-ICP

The integrated care pathway tool for the myotonic dystrophy type 1 clientele (DM1-ICP) was developed to be used within the nursing practice for individuals with a late or adult DM1 phenotype. This tool can be used for initial evaluation and regular follow-up at neuromuscular disease clinics and home-care visits. The DM1-ICP is a tool that was designed to support the nursing practice but does not replace nurse autonomy or critical judgment. It was developed to respond to several needs, including:

1) The need to review the services offered to ensure better use of professional resources;
2) The need to integrate evidence-based data within the nursing practice.

A brief description follows of the several stages that were completed allowing for the creation of DM1-ICP:

- *Critical review of literature:* Bibliographic research of the topic from 1980-2008 was completed in the following databases: PubMed, CINAHL, PsychINFO, PsychFIRST. The following key words in French and English were used: myotonic dystrophy, neuromuscular disorder, neuromuscular disease, Steinert disease, Steinert disorder, and Steinert syndrome. The articles concerning the following subjects were excluded: myotonic dystrophy type 2, chromosome mapping, genetic markers, linkage, genetics polymorphism, mutation and cells.

- A total of 4,212 articles were identified, of which 847 were pertinent to the current project. The systematic revision of the articles was completed using the Scottish Intercollegiate Guidelines Network (SIGN) method[1]. During the article-review process, it was observed that the majority of the articles were of level IV, consisting of either expert opinion or were non-analytical. There were no recommendations at the I-II-III levels (randomized studies).

The identification of the systemic and social concerns about DM1 and the elaboration of recommendations for management have been done according to the summary of the opinions of experts in DM1 through a Delphi process. We also used data gathered in the context of a Canadian Institute of Health Research (CIHR)-funded project entitled "Consequences of Neuromuscular Genetic Disorders: Determinants of Disabilities, Social participation, and Quality of Life in DM1". This project has been designed to improve the understanding of the natural history of DM1 and to identify significant covariates of social participation, well-being and quality of life for affected patients and their families in order to

promote comprehensive health supervision strategies. Some results of this project have already been published and others are discussed in the present document under the heading "DM1-CIHR project". See «Health supervision and anticipatory guidance in adult myotonic dystrophy type 1" reported in Neuromuscular Disorders [2] for the complete methodological approach.

## CONDITIONS OF USE

Considering that DM1-ICP is an exhaustive evaluation tool that helps assess the health state of a person with DM1, the use of different competencies specific to the nursing profession is required. During the development of the DM1-ICP, each one of its elements were evaluated with regards to what a nurse could do to ensure better health management and follow-up of people with DM1.

**The recommendations should be reviewed in each country in order to reflect current practices, professional legislation and organization of care.**

> In Quebec, legislative framework recognizes the role and responsibilities of nurses as:
>
> ...evaluating a person's state of health, determining and assuring the provision of a nursing care-and-treatment plan, unsparingly providing nursing and medical care and treatments with the goal of maintaining and regaining health, preventing disease and providing palliative care.[3]

In addition, the same legislative framework recognizes that nurses can carry out the following reserved activities that are in line with the activities stipulated in DM1-ICP:

1. Evaluate the physical and mental condition of a symptomatic person.
2. Carry out the clinical surveillance of the physical condition of those whose state of health presents risks, including monitoring and adjusting the nurse therapeutic plan.
3. Initiate diagnostic and therapeutic measures, as collectively prescribed.
4. Conduct diagnostic tests and examinations, as collectively prescribed.
5. Provide and adjust medical treatments, as collectively prescribed.
6. Provide regular nurse follow-ups to those presenting complex health problems.

The elements of evaluation, intervention and reference, which refer to the following competencies, are part of the case management approach in nursing[4]:

- Know and apply the principles of the partnership: recognize the person's expectations and potential, suggest pertinent approaches, respect the person's right to make health choices and decisions, share responsibilities, accompany and provide support.
- Be capable of carrying out an evaluation of the person's health situation: evaluate the person's physical and mental state and surrounding physical and social environment, verify personal resources, evaluate risks, assess learning needs, etc.
- Evaluate personal state by using the results of the diagnostic tests and consultations and by proceeding with the necessary diagnostic tests as per a collective prescription.
- Determine what clinical follow-up is required and adjust it as needed as per the evolution of the person's health situation and efficiency of the interventions.
- Help the person use and increase the personal repertoire of resources to be able to maintain and improve health and well-being or prevent sickness, accidents, social problems, mainly by adopting healthy habits.
- Establish scanning, surveillance and follow-up procedures, in risk situations and eliminate, if possible, risk factors linked to the physical and social environment of the person.
- Relay to other personnel, when needed and at opportune times, information related to the health of the person. Determine the nature of the information based on the situation and assure the coordination and follow-up of the consultation, if the need arises.
- Coordinate interventions of the interdisciplinary team, while taking into consideration the health situation of the person, particularly when conducting systematic follow-ups with clientele.

More nursing competencies are hereinafter referenced[4]:

- Know the human anatomy, physiology, biochemistry, microbiology and their relations with the disease and the different dysfunctions.
- Know the medical and surgical treatments in addition to their relation with explanatory mechanisms.
- Know and apply the underlying scientific principles of care interventions based on diagnostic, medical and surgical treatments.
- Know pharmacology and apply principles of medicinal administration: action, side effects, undesirable effects, secondary effects, interaction, and pharmacovigilance.
- Know the diagnostic tests and how they relate to the evolution of the state of health and explanatory mechanisms.
- Know the fundamentals of developmental psychology, social psychology, health and family sociology.
- Know the different health experiences lived by the individuals.
- Know the cultural differences with regards to health experiences.
- Know and apply teaching principles.

- Know and apply the mechanisms that permit respect of the clientele's legal rights.
- Know and apply the principles related to inter-professional collaboration.
- Know the tools and technology used for evaluations, clinical surveillance, care and treatments and how to operate them.
- Verify the person's response to interventions.
- Plan care activities based on individual needs, expectations, and resources, in collaboration with the support network.
- Help individuals adapt to inherent change in personal growth, development, and different stages of life and stressful events.
- Inform and support individuals, allowing them to make informed health decisions.
- Collaborate with various personnel in the planning and implementation of functional rehabilitation programs.
- Help individuals exploit their potential and recover their autonomy in various aspects of life such as mobility, nutrition, security, etc.

A final condition of use concerns collective prescriptions that the nurse can allocate as per new legislative code (Bill 90).[5] Before the nurse can begin diagnostic exams and giving referrals as foreseen in DM1-ICP, the organization must approve the instances where collective prescriptions are required.

## PORTRAIT OF MYOTONIC DYSTROPHY TYPE 1

Myotonic dystrophy type 1 (DM1) is the most common type of muscular dystrophy in adults.[6] The prevalence of DM1 varies from 2.1 to 14.3 cases per 100,000 worldwide but reaches 189 cases per 100,000 in the Saguenay-Lac-Saint-Jean (SLSJ) region of Quebec.[7, 8] DM1 is an autosomal dominant disease caused by unstable trinucleotide repeat expansion of the cytosine-thymine-guanine (CTG)n located on the 19q13.3 chromosome.[9] The severity of the disease is globally correlated to the number of CTG repeats on the gene.[10] DM1 is characterized by the abnormalities present in several systems including the muscular, respiratory, cardiac, endocrine, ocular, and central nervous systems.[6, 11] Typically, symptoms become obvious during mid-life however first signs may be detectable within the first decade of life.[6] DM1 affects several aspects of life including: 1) a reduction in life expectancy with only 12% of people living longer than 65;[12, 13] 2) compromised social participation in several spheres as only a small portion of DM1 individuals maintain an active and satisfying social life;[14, 15] and 3) a living environment characterised by poverty, social exclusion and an under-developed health management approach.[16-21] The variable clinical profile and the substantial amount of simultaneous co-morbidities that characterize DM1, which can often be considered as a model of premature aging, render management of the disease a constant challenge.[21, 22] The DM1 population requires a progressively established nursing rehabilitation approach consisting of the timely presence of nurses, as defined by Booth and Jester (2007).[23]

The description and portrayed manifestations of DM1 must reflect variability of the disease. Consequently, DM1 is classified into four clinical phenotypes: congenital, childhood, adult and late.[24] The phenotypes of DM1 are determined by the age of onset and by the number of CTG repeats that occur. DM1 is caused by an abnormal amplification of a CTG nucleotide triplet on the 19q13.3 chromosome.[9, 25, 26] For non-affected individuals, the number of triplet repeats is between 5 and 35 whereas for individuals with DM1, between 50 and several thousand CTG repeats have been observed.[27] The congenital and childhood phenotype (1,000 repeats or more), the adult phenotype (between 100 and 1,000 repeats) and the late phenotype (between 50 and 150 repeats) are terms used by the medical community.[10, 27] In the congenital phenotype, first manifestations present at birth consist of hypotonia, delayed motor development, and delayed mental development; manifestations in other organs occur later in the disease course.[6, 28] The main characteristics of the childhood phenotype which begin to appear between the ages of one and 10 are: mild facial weakness, hypernasality, presence of myotonia and learning difficulties.[28] The congenital and childhood phenotypes have distinct clinical portraits and are the most severe. People with the adult phenotype begin showing symptoms between the ages of 10 and 40 and demonstrate a significant variety

of problems consisting of progressive loss of muscle strength and presence of myotonia. The adult phenotype is the most frequent form of DM1.[6] For those with the late phenotype which manifests at around the age of 40, often minimal signs and symptoms are present and include cataracts and mild myotonia with little muscular weakness.[29] The present document is principally interested in the adult and late phenotypes. Childhood and congenital phenotypes have completely different clinical presentations and will not be discussed further in this document.

SECTION 4

## CURRENT STATUS OF PROVISION OF CARE TO DM1 CLIENTELE

Neuromuscular diseases are part of a broad range of diseases that may or may not be hereditary. Among the hereditary diseases, there are muscular dystrophies, spinal atrophies, metabolic myopathies and myotonias.[30] The services offered to people with neuromuscular diseases are generally described as deficient and inadequate.[31-34] A report published in England in 2007 indicates that the amount of follow-up offered to people with neuromuscular disorders differs from region to region, that the services vary and are vulnerable due to changes in staff and that the patients do not receive the multidisciplinary care and services required for their condition.[33]

In Canada, the services are often organised at specialised neuromuscular clinics, which are usually interdisciplinary and with variable service organization. Several partners are also involved in the follow-up of the DM1 clientele including family physicians, local community services, rehabilitation centres and the local chapters of Muscular Dystrophy Canada. The role and responsibilities of nurses vary from one clinic to another. To our knowledge, no report or study has focused on the organisation of treatment and health services offered to people with DM1 across Canada.

The Clinic of Neuromuscular Diseases at the *Centre de santé et de services sociaux de Jonquiere (CSSS Jonquière)*, Quebec, Canada proposes a model to organise services around a nursing case manager who is responsible for screening the needs of the clientele, evaluating the identified needs, engaging in a personalised referral process of the different health professionals and community services available in addition to providing specialised expert medical care. One of the responsibilities of the nursing case manager is to use an integrated health management tool (DM1-ICP) when screening the needs of people with DM1. This tool was developed within a research project involving researchers from the *Groupe de recherche interdisciplinaire sur les maladies neuromusculaires (GRIMN)*, the interdisciplinary research group of neuromuscular diseases, and nurses from the clinic.

SECTION 5

## EVALUATION PROCEDURES

This section presents the evaluation of various impairment and disabilities can be observed in the various organic systems in people with DM1. Firstly, the evaluation of the metabolic and endocrine, visual, respiratory, cardiovascular, gastrointestinal, muscular, central nervous and reproductive systems are presented. Then, the evaluation of risk factors as well social participation aspects is presented.

SECTION 5

## EVALUATION OF THE METABOLIC AND ENDOCRINE SYSTEMS

The human body is regulated by the nervous system and by the endocrine system. The nervous system acts rapidly and briefly through nerve impulses whereas the endocrine system acts slowly and over a longer period of time through hormones.[35] The main endocrine glands are: the pineal gland, hypothalamus, pituitary gland, thyroid and parathyroid glands, thymus, suprarenal glands, pancreas and the gonads.

The most widespread endrocrine abnormalities in DM1 are testicular atrophy and an alteration of insulin metabolism.[6] Several studies on the endocrine system indicate gender differences: men are more severely affected than women. Reasons for the endocrine perturbations and gender differences are not clear and may implicate several different mechanisms including the severity of the genetic defect (CTGn), which can in turn, affect response, specific tissue metabolism and hormone production.[36]

## METABOLIC AND ENDOCRINE SYSTEMS

### ELEMENT OF SURVEILLANCE
Diabetes

| DIABETES | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Diabetes: Fasting Glucose | Normal ☐<br>Abnormal ☐<br>Unknown ☐ | At baseline and repeat every three years.<br>Refer patient to his general practitioner if results are abnormal according to national guidelines. |

As much as glucose intolerance and insulin resistance are known characteristics of DM1, diabetes is relatively infrequent, with a prevalence between 0 and 6.7%, and that regardless of glucose intolerance levels at around 20%.[6, 36]

**Glucose intolerance**

People with DM1 do not seem to share the same glucose metabolism profile as the general population. In the DM1 population, a high level of glucose intolerance has been observed without there being any elevated prevalence of diabetes, which would be expected based on observations of the general public.

**Insulin resistance**

People with DM1 often have normal basal level of insulin in their blood. However, excessive releasing of insulin is observed when there is an increase in glucose, indicating a compensatory response of ß cells to insensitivity in insulin tissues.[37, 38] There is no evidence that the insulin produced by people with DM1 is abnormal.[39]

Insulin resistance would be a secondary side effect linked to the reduction of the insulin receptor RNA. A study indicated that the RNA level and the insulin receptor protein of people with DM1 were reduced by 50% when compared to a control group [40] The effect of trinucleotide CTG repeats on RNA translation not only involve DMPK (Myotonic Dystrophy Protein Kinase) RNA but also other RNA such as the RNA of the insulin receptor.[40] In the DM1 cells, the RNA code muted by DMPK accumulates in the nucleus and links with numerous proteins to form an abnormal intranuclear structure. Numerous functional modifications follow in the cell, such as alternative splicing of insulin receptor RNA.[41]

Insulin resistance observed in DM1 clientele is a peripheral type;[42] people with DM1 still have normal glucose levels , even though insulin resistance should lead to hyperglycemia, the organism adapts by increasing insulin secretion.

**NURSING INTERVENTIONS**

There is no way to screen for insulin resistance because no simple and reliable method exists. Instead, glucose intolerance is tested by using an oral glucose tolerance test (2 hours). When screening for pre-diabetes, especially within the context of metabolic syndrome (see dyslipidemia section), it is possible to determine which patients would benefit from modifying their lifestyle related to cardio-vascular risk factors.[43] It has been demonstrated that lifestyle changes are very efficient at delaying and preventing the onset of diabetes in people with glucose intolerance.[44, 45] If the person presents additional risks factors related to diabetes, the evaluation frequency of glucose-levels while fasting may increase or become more important.[46]

One of the nurse's first responsibilities is to test serum glucose while fasting and analyze the result. If the result is abnormal, the nurse must follow national recommendation on diabetes management and refer the client to his general practitioner for a complete investigation.

## METABOLIC AND ENDOCRINE SYSTEMS

### ELEMENT OF SURVEILLANCE
Hypothyroidism

| HYPOTHYROIDISM | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Hypothyroidism: TSH. | Normal ☐<br>Abnormal ☐<br>Unknown ☐ | At baseline and repeat every three years.<br>Refer patient to his general practitioner if results are abnormal. |

The problems with thyroid functioning in the DM1 population shows a prevalence similar to the general population.[6, 36] Some clinical characteristics of DM1 such as myotonia, apathy, fatigue, somnolence, excessive daytime sleepiness and general sluggishness may be initially mistaken for hypothyroidism or, in the course of the disease, may cover up such a thyroid disturbance.

**NURSING INTERVENTIONS**

In the presence of abnormal results, the nurse refers the patient to his general practitioner and afterwards, must follow-up to ensure adequate health management. There is no DM1 specific intervention. Follow national clinical guideline for hypothyroidism.[36]

## METABOLIC AND ENDROCRINE SYSTEMS

### ELEMENT OF SURVEILLANCE
Hypogonadism

| HYPOGONADISM | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Hypogonadism: Total testosterone, FSH, LH.<br>*Men only.* | Normal ☐<br>Abnormal ☐<br>Unknown ☐ | At baseline only, except if suggestive symptoms of hypogonadism.<br>Refer patient to his general practitioner if results are abnormal. |

Hypogonadism is present in 60% to 90% of men with DM1 and it is of primary type as it originated from a dysfunction of the gonads due to a secondary overproduction of gonadotropin. The level of blood testosterone uniformly diminishes with high levels of follicle-stimulating hormone (FSH) and an increase in luteinizing hormone (LH).[6, 36] In women, there is little evidence of hypogonadism or any other form of gonad dysfunction.[6]

Hypogonadism and the local degeneration of smooth muscle tissue could be implicated in erectile dysfunction frequently observed in men with DM1 (see the reproductive system section). Fertility remains normal.[6] The symptoms associated with reduced testosterone in men can include: dry skin, fatigue, loss of muscle/atrophy, low libido, erectile dysfunction and weight gain.[6] Administration of testosterone has not been shown to be an effective treatment for loss of muscular function for the DM1 population.[36]

**NURSING INTERVENTIONS**

Refer patient to his general practitioner if results are abnormal. There is no DM1 specific intervention. Follow national clinical guideline for hypogonadism.

## METABOLIC AND ENDOCRINE SYSTEMS

### ELEMENT OF SURVEILLANCE
Dyslipidemia

| DYSLIPIDEMIA | | |
|---|---|---|
| **Evaluation** | | **Intervention/Reference Criteria** |
| Dyslipidemia: Level of blood lipids. (Triglycerides, total cholesterol, LDL, HDL, apolipoprotein B) | Normal ☐<br>Abnormal ☐<br>Unknown ☐ | At baseline and repeat every 3 years.<br>Refer patient to his general practitioner if results are abnormal.<br>Refer patient to the lipid clinic as needed. |

Several reports confirm elevated levels of triglycerides and cholesterol while fasting in the blood plasma of people with DM1.[36, 47] However, the high cardiac mortality rate observed in DM1 [13] is rarely linked to ischemic heart disease that can occur from dyslipidemia.[48]

Several elements suggest that DM1 progresses towards the expression of a classic variant of metabolic syndrome.[36] Metabolic syndrome exists when an individual shows[49] central (abdominal) obesity measured by the circumference of the waist (men >102 cm; women >88 cm). Moreover, if the body mass index is >30 kg/m$^2$, central obesity can be assumed. Central obesity must be accompanied by at least two of the four following factors: 1) raised triglycerides (≥150 mg/dl (1.7 mmol/L) or specific treatment of this lipid abnormality), 2) reduction HDL cholesterol (men <40 mg/dl; women <50 mg/dl or specific treatment of this lipid abnormality), 3) raised blood pressure (Systolic BP ≥ 130 or diastolic BP ≥ 85 mm Hg or treatment for previously diagnosed), and/or 4) raised fasting plasma glucose (≥ 100 mg/dl (5.6 mmol/L) or previously diagnosed type 2 diabetes). However, blood pressure issues in DM1 is hypotension instead of hypertension as often seen in general population[36] and type 2 diabetes is infrequent [6, 36]. Regardless of the observed abnormalities, people with DM1 do not have increased incidence of cardiovascular disease or diabetes.[50]

**NURSING INTERVENTIONS**

The nurse must be alert for symptoms that may result in dyslipidemia especially with men over 40 years old, with women over 50 years old or postmenopausal and with people who have an elevated-risk profile (they have at least one of the following elements: hypertension, tabagism, obesity, family history, early cardiovascular problems and/or physiological manifestations of hyperlipidemia).[51] Interdisciplinary management must be established by the nurse.

## METABOLIC AND ENDOCRINE SYSTEMS

### ELEMENT OF SURVEILLANCE
AST, ALT, Bilirubin, Gamma-GT, CK

| CHRONIC INCREASE OF AST, ALT, BILIRUBIN, GAMMA-GT, CK | | |
|---|---|---|
| **Evaluation** | | **Reference/intervention Criteria** |
| Chronic elevation of AST, ALT, Bilirubin, Gamma-GT | Normal ☐<br>Abnormal ☐<br>Unknown ☐ | At baseline only. |

The prevalence of elevated levels of CK, AST, ALT and bilirubin are unknown. However, elevation of the serum gamma-glutamyltranspeptidase (gamma-GT) has been observed in a small sample of people with DM1, of whom the average elevation was five times higher than accepted values.[52]

Gamma-GT are enzymes that participate in the metabolism of amino acids, which are base elements of proteins. Gamma-GT are present in the liver, the kidneys and the pancreas. Elevation of serum gamma-GT in people with DM1 is due to a minor malfunction of the liver.[52.] Most people who have elevated gamma-GT also present one or many more abnormal results in terms of hepatic function (AST, ALT and bilirubin) however medically significant hepatic ailments are not present. Considering that most clinicians are unfamiliar with minor chronic liver malfunctions in DM1, the purpose of the hepatic function tests at baseline during a routine check-up is to prevent useless and invasive future investigations.

**NURSING INTERVENTIONS**

Refer patient to his general practitioner if the results are abnormal. There is no DM1 specific intervention. Follow national clinical guideline.

## EVALUATION OF THE VISUAL SYSTEM

Several manifestations in the visual system have been observed in DM1. However, there is no systematic management of these manifestations and these are beyond the scope of the present document. The impairment can include eyelid hypotonia[53-56], extraocular myotonia[57], miosis [56], pigment change in the macula, abnormalities affecting the ocular pursuit[57-62], ocular motricity, weakness[56] and divergent strabismus[63, 64].

## VISUAL SYSTEM

### ELEMENT OF SURVEILLANCE
Ptosis

| PTOSIS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Presence of ptosis?<br>If yes, does it interfere with your vision? | Y ☐ N ☐<br>Y ☐ N ☐ | Specify: _____<br>If yes, refer to an ophthalmologist. |

People with DM1 rarely say they suffer from oculomotor symptoms other than myogenic ptosis.[60] Palpebral ptosis, abnormal drooping of the upper lid, is a frequent condition that is most often bilateral, symmetrical and generally severe.[65, 66] It is caused by partial or total reduction in elevator muscle function of the upper eyelid. Ptosis can be more or less severe, depending on whether it hinders the visual axis and impedes visual functioning.[65]

**NURSING INTERVENTIONS**

If ptosis is present and impeding vision, refer to an ophthalmologist. The surgical treatment of ptosis in DM1 can be possible under certain circumstances. The ophthalmologist must suggest this intervention only with the goal of improving functional vision.[66] In order to avoid complications, ptosis surgery must only occur when the visual axis is hindered.[65] In the case of surgical treatment, it is recommended to review the risks associated with anaesthesia.

## VISUAL SYSTEM

### ELEMENT OF SURVEILLANCE
Cataracts

| CATARACTS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have cataracts? | Y☐ N☐ | If yes, frequency of follow-up with the optometrist or ophthalmologist: _____ |
| Surgical treatment of cataracts? | Y☐ N☐ | Date: _____ |
| If no cataracts:<br>• Do you have blurred vision? | Y☐ N☐ | • If yes to either of the questions and if the visual acuity test is inferior to 20/50, refer to an optometrist then to the ophtalmologist if needed. Refer to physician for driving assessment. (report to driving authority if the patients still drive) |
| • Are you sensitive to glare? | Y☐ N☐ | |
| Visual acuity test<br>*(Rosenbaum Pocket Vision Screener)* | Result: _____<br>Normal ☐<br>Abnormal ☐ | If appropriate, ask patient to wear his glasses for the visual acuity test:<br>• The card must be held in a manner for it to clearly be seen, at a distance of 14 inches from the patient.<br>• Ask the patient to read each line starting from the top (20/800). |
| • If the patient wears glasses, when were they last adjusted? | | _____<br>_____<br>_____ |

In DM1, it has been recognized for a long time that cataracts are very frequent, quasi constant, inevitable, and appear early-on in the progression of the disease.[53-56, 67, 68] Cataracts can be, in certain cases, the only sign of the disease. A study has shown that 91% of people with DM1 have bilateral, subcapsular cataracts.[69] Regardless of the fact that the majority of people with DM1 maintain functional visual acuity, a study has reported that nearly 27% of this majority show visual acuity inferior to 20/60 due to cataracts.[55] The pathological process responsible for cataracts in DM1 is determined by the genetic abnormality (CTGn).[70]

It is recognized that reduction of visual acuity in DM1 is a main consequence of the progression of cataracts.[55] Surgical removal of cataracts is possible when the cataract has reached maturity. However, surgical complications, other than those related to anaesthesia, have been described for people with DM1.[71-73] The reoccurrence of a second loss of transparency, among other complications, has been

reported as frequent following cataract surgery[74], and it seems that people with DM1 are predisposed to this complication[72].

**NURSING INTERVENTIONS**

The *Rosenbaum Pocket Vision Screener* is the chart most often used worldwide to measure visual acuity.[75] The person must wear his glasses for the test, if applicable. The chart must be held at a distance of approximately 14 inches from the eyes. In the case of surgical treatment for cataracts, it is recommended to review the risks associated with anaesthesia with the patient. In case of reduced vision below 20/50 referred to the general practitioner for driving evaluation.

# EVALUATION OF THE RESPIRATORY SYSTEM

Several complications of the respiratory system can occur in people with DM1 such as chronic ventilatory failure, pneumonia, and sleep related breathing disorder. Respiratory impairment is the leading cause of mortality in DM1 patients.[12, 13] Another issue of the respiratory system concerns anesthetic risks, with perioperative complications being common in DM1[76].

## RESPIRATORY SYSTEM

## ELEMENT OF SURVEILLANCE
Chronic respiratory failure

| Chronic Respiratory Failure | |
|---|---|
| **Evaluation** | **Reference/Intervention Criteria** |
| 1. Not troubled by breathlessness except on strenuous exercise.<br>2. Short of breath when hurrying or walking up a slight hill.<br>3. Walks slower than contemporaries on level because of breathlessness, or has to stop for breath when walking at own pace.<br>4. Stops for breath after walking about 100 m or after a few minutes on level ground.<br>5. Too breathless to leave the house, or breathless when dressing or undressing.<br><br>Respiratory assistance: | If dyspnea grade 3/5 or more, proceed with a pulmonary X-ray and respiratory function tests and refer patient to his general practitioner.<br><br>BiPAP ☐ Adequate Use Y ☐ N ☐<br>$O_2$ ☐<br>Other ☐ |

Chronic respiratory failure is the primary cause of mortality and morbidity for people with DM1, whose life expectancy is often reduced.[13, 77] Just like in the case of pneumonia, inspiration and expiration muscles are both affected and are in part responsible for the breathing difficulties associated with DM1. It is also suspected that the central nervous system and respiratory functions are primarily affected.[12] Once proximal muscle weakness is apparent, respiratory muscle capacity and forced vital capacity decline considerably.[78]

**NURSING INTERVENTIONS**

The *Medical Research Council Dyspnea Scale* (MRCDS) evaluates the degree of breathlessness (dyspnea) of a patient and the repercussions it may have of the daily activities of the patient.[79-81] Evaluation for dyspnea in DM1 is complicated by the presence of excessive daytime sleepiness and fatigue. Moreover, the questionnaires used to evaluate dyspnea have not been developed for DM1 and the questions are often inappropriate for the DM1 population. Nevertheless, the clinician must suspect respiratory complications each time dyspnea is present.

If dyspnea grade 3/5 (MRCDS) or more, proceed with a chest X-ray and pulmonary function tests and refer patient to general practitioner. The nurse can assess the patients' experience of fatigue and its

contributing factors, monitor the patients' level of fatigue and their use of existing strategies to manage it, educate patients in the application of strategies to manage fatigue (e.g. physical activity, energy conservation, relaxation, diversion, sleep promotion and proper nutrition) and coach patients in the selection and performance of strategies to manage fatigue.[82] Nonpharmacological nursing interventions for the management of patient fatigue: a literature review. *Journal of Clinical Nursing*.). In addition, the nurse may consider a referral to the occupational therapist to identify and develop modifications to their daily activities in order to reduce fatigue.[83]

## RESPIRATORY SYSTEM

## ELEMENT OF SURVEILLANCE
Pneumonia

| PNEUMONIA | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| How many pulmonary infections treated with antibiotics have you had during the last 6 months? | Number _____ | If more than one infection, refer patient to his general practitioner. |

Recurrent pulmonary infections resulting from aspirations are frequent in DM1. Pneumonia is described as the primary cause of death in two studies regrouping 70[12] and 75[13] people with DM1. In the studies, the mortality rate was established at 31% and 28% respectively. When both the inspiratory and expiratory muscles (the diaphragm and the intercostal muscles) are affected, the capacity to cough is reduced and there is an alteration in coughing reflexes.[6, 84]

**NURSING INTERVENTIONS**

Cough assist devices are used in some clinics throughout the world, however to date, no study that evaluates their efficiency exists. If more than one infection, refer patient to his general practitioner.

## RESPIRATORY SYSTEM

## ELEMENT OF SURVEILLANCE
Vaccination

| VACCINATION | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Did you receive the influenza vaccine last year? | Y ☐ N ☐ | Annually refer all patients for the influenza vaccine and explain the importance of the vaccination. |
| Have you ever received the Pneumovax vaccine? | Y ☐ N ☐ | Date of last Pneumovax vaccine: _____<br><br>Once in a life time Pneumovax vacine for most patients; those with chronic respiratory disease, spenectomy, asplenia or terminal renal failure will require a five-year renewal of this vaccine. |

Annual vaccination against influenza is recommended for all people with DM1[2]. In DM1, immunization against the influenza virus and pneumococcal bacteria is particularly important for people who have reduced lung volume or proximal muscle weakness. Moreover, people who have difficulty expelling their respiratory secretions and/or at increased risk of aspiration must receive particular attention. The Pneumovax vaccine is also recommended for the at-risk individuals.

**NURSE INTERVENTIONS**

Refer for vaccination.

# RESPIRATORY SYSTEM

## ELEMENT OF SURVEILLANCE
Risks associated with anaesthesia

| RISKS ASSOCIATED WITH ANESTHESIA | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Have you been informed of the risks associated with anesthesia in DM1? | Y ☐ N ☐ | Validate the person's comprehension of the risks associated with anesthesia.<br>If understanding is inadequate, proceed to information using the corresponding section of the Myotonic Dystrophy Foundation. Moreover, provide the DMC alert card, to be inserted in the person's wallet or purse, in case of accident or emergency.<br>Provide to the patient the factsheet for professionals. |

Several studies report elevated risk of complications during or after general anaesthesia.[76, 85, 86] A retrospective study conducted with a large population sample showed a prevalence of anaesthetic complications at around 10%.[76] The majority of complications observed were pulmonary. People with DM1 are particularly at risk during and after general anaesthesia because their muscular, cardiovascular and respiratory systems are affected by the disease. Local or epidural anaesthesia should be considered depending on the case. Primary anaesthetic considerations in DM1 include myotonia, temporomandibular subluxation and intubation difficulties, cardiac arrhythmia, heart failure, hypotension, respiratory failure, somnolence, gastroesophageal complications and risk of pulmonary aspiration.[87] Excellent results can be obtained by performing a thorough preoperative evaluation, anticipating and minimizing complications through selection of the most appropriate anaesthetic and surgical techniques. Close surveillance must be assured during the postoperative period.[87]

### NURSING INTERVENTIONS

Summary of the practical suggestions for the anesthetic management of a DM1 patient (from provision of an Alert Card [2] and Myotonic Dystrophy Foundation, Toolkit; http://www.myotonic.org):

- Perform an extensive preoperative evaluation. Organize a multi-disciplinary medical team.
- Use regional anesthesia when appropriate.
- Be cautious with premedications (benzodiazepines and opioids).

- Keep the patient warm.
- Consider applying defibrillator/pacer pads.
- On induction, be aware of the high likelihood of aspiration and other airway complications. Avoid succinylcholine when possible.
- Adhere to strict extubation criteria. Given the effects DM has on the pulmonary system, anticipate the need for supportive mechanical ventilation until extubation criteria are met.
- Plan for the continuous SpO2 and EKG monitoring postoperatively.
- Manage postoperative pain with NSAIDs, regional techniques, and acetaminophen when appropriate. Use opioids with extreme caution.
- Encourage aggressive pulmonary toileting postoperatively.

# EVALUATION OF THE CARDIOVASCULAR SYSTEM

Complications of the cardiovascular system in DM1 refer to conduction defects and arrhythmia, blood pressure dysregulation, and cardiomyopathy [88].

## CARDIOVASCULAR SYSTEM

### ELEMENT OF SURVEILLANCE
Cardiac conduction defects and arrhythmias

| CARDIAC CONDUCTION DISTURBANCES AND ARRHYTHMIAS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have a pacemaker? | Y ☐<br><br>N ☐ | If yes, year of implantation: _____<br>• Be sure that the patient with a pacemaker has a regular follow-up every six months at a pacemaker clinic.<br>**If no pacemaker:**<br>Is there family history of sudden cardiac death?<br>• If yes, proceed with an ECG and a Holter than refer to a cardiologist according to results. |
| **ECG:**<br>• Date of last ECG<br><br>• Result of last ECG<br><br>_____<br>_____<br>_____<br>_____ | _____<br><br>Normal ☐<br>Abnormal ☐ | Refer to a cardiologist in the following situations:<br>1. Atrial fibrillation or flutter (verify if anticoagulants have been taken)<br>2. Second-degree or third-degree AV block<br>3. Junctional cardiac rhythm<br>4. PR interval>200 msec<br>5. QRS interval >105ms<br>6. Right bundle-branch block with a hemiblock, in particular if associated with a AV block<br>7. Symptomatic sinus bradycardia<br>8. Unexplained fainting or loss of consciousness |
| Over the last year, have you suffered from:<br>• Loss of consciousness<br>• Dyspnea<br>• Orthopnea<br>• Edema (swelling) of lower limbs<br>• Palpitations<br>• Dizziness<br>• Lipothymia (faintness)<br><br>Vital signs:<br>• Heart beat (pulse)<br><br>• Cardiac rhythm | Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐<br><br>Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐<br><br>_____ beats/minute<br><br>Regular ☐<br>Irregular ☐ | Note the frequency of each symptom:<br>_____<br>_____<br>_____<br>_____<br>_____<br>_____<br>_____<br>_____<br>_____<br>_____<br>_____<br><br>Proceed with a Holter and refer to a cardiologist if symptoms of sinus bradycardia:<br>1. If unexplained faint or loss of conciousness, proceed with an ECG and refer patient to a cardiologist.<br>2. If signs or symptoms of cardiac insufficiency (dyspnea, orthopnea, lower limbs edema) proceed to a pulmonary X-ray, an echocardiography and refer patient to a cardiologist if results are abnormal.<br>3. If palpitations, proceed with an ECG and a Holter and refer patient to a cardiologist if results are abnormal. |

Several abnormalities of the cardiac system have been observed in people with DM1.[89, 90] The deterioration of the cardiac system is generally slow, but remains unpredictable.56,[91] It has been observed that approximately 65% of the DM1 population present with an abnormal ECG.[92, 93] Regardless of the fact that few people with DM1 complain of cardiac symptoms, the abnormalities of this system are the second leading cause of death in DM1.[94] Cardiac complications were the leading cause of death in 29% of a first series cases[12] and 20% in a second series of 367 cases during a 10-year period[13]. People presenting electrocardiographic abnormalities are more susceptible to systolic dysfunction and heart failure.[95]

Cardiovascular system abnormalities can be divided into two categories: cardiac conduction defects and arrhythmias. Cardiac conduction defects are pathophysiological states that can be present in people with DM1 and are responsible for putting them more at risk for cardiac arrhythmia. Any part of the cardiac conduction system can be affected, but the His-Purkinje system is most often involved.[92] The most common conduction defects are: first degree atrial-ventricular block (AV) with prolonged PR interval, left anterior hemiblock, left or right bundle branch block, and prolongation of the QT interval.[90] Several types of arrhythmias have equally been observed in people with DM1.[94] These people can present with supraventricular arrhythmias and ventricular tachyarrhythmias, however the latter is most common. The most common arrhythmias are atrial flutter or atrial fibrillation observed in more than 25% of people with DM1 [91], and both may be intermittent or chronic. Ventricular arrhythmias are also frequent, especially ventricular tachycardia.[91]

Even though the explanatory mechanisms of cardiac problems associated with DM1 are not completely elucidated, histological changes have been well documented.[89] Endomyocardial biopsies and post-mortem studies have shown several degrees of non-specified modifications, such as progressive interstitial fibrosis, fatty infiltration in conjunctive tissues of the conduction system and hypertrophy of myocardiocytes (myocardial muscle cells).[48, 91] Fatty infiltration of the myocardium is always present in people with a more severe form of DM1 and is associated with more advanced cardiac conduction defects.[96] Cardiac tissue fibrosis offers a favorable environment for the development of cardiac conduction defects and electrical ectopic activity.[48] The histological modifications (fibroid degeneration of conduction tissue) can first result in supraventricular cardiac conduction disturbances.

Minor cardiac conduction disturbances are often observed in the first stages of DM1. Such initial cardiac symptoms are most often manifested though asymptomatic abnormalities in the ECG, such as prolongation of PR or QRS intervals. A progression towards more severe cardiac conduction

disturbances can also be asymptomatic, but they may also move towards a bradycardia or a symptomatic tachycardia manifested in the form of syncope or cardiac arrest (sudden death) [91]. Some potential problems such as first degree AV block, second or third degree AV block and second or third degree AV block can be observed in patients DM1 [97].Sudden death of cardiac origin is difficult to predict in people with DM1. Recently, some severe cardiac abnormalities have been identified as risk factors for sudden death within the DM1 population: 1) cardiac rhythms other than sinus; 2) a PR interval ≥ 240 msec; 3) a QRS interval ≥ 120 msec; 4) AV block of the second or third degree; and 5) supraventricular tachyarrhythmia such as atrial fibrillation.[95] Another study that followed 428 people with DM1 over a period of more than ten years showed that a PR interval of more than 200 msec and a corrected QT interval more than 459 msec were related to sudden death or the implantation of a pacemaker.[98] Recently, a study highlighted that addition of each year of age, being a male and severity of muscular impairment are associated with the development of cardiac arrhythmias, but remain weak as risk indicators associated with fatal arrhythmias.[99]

**NURSING INTERVENTIONS**

Several people with DM1 have pacemakers. Follow-up at the pacemaker clinic is essential. Follow-up every six months as required by national and international clinical directives.[100, 101]

For people with DM1 who do not have pacemaker, an annual ECG is an international directive.[90] [2]Recent norms in practice prescribe conducting further investigation with people who have DM1, of whom at least one family member suffered sudden death of cardiac origin.[90] Referral to a cardiologist for investigation and ongoing care will vary from one clinic to another and is still a matter of debate even among experts. From the consensus of several experts, proposed referral criteria of DM1 patients to a cardiologist are listed in the corresponding table. The recommended frequency of electrocardiogram testing for all people with DM1 is at least once a year.[90]

Even though they remain infrequent, the symptoms that may suggest cardiac system abnormalities have been described in several studies and should be addressed properly.[6, 34, 90] The measure of cardiac frequency allows obtaining information on the following parameters: the frequency, cardiac rhythm, and pulse amplitude. Refer to cardiologist if abnormal results.

## CARDIOVASCULAR SYSTEM

## ELEMENT OF SURVEILLANCE

Hyper or hypotension

| HYPER OR HYPOTENSION | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| • Blood pressure (BP) | ____/____<br>mm Hg | **Hypotension**<br>1- If hypotension: verify BP on both arms and refer back to cardiac symptomology.<br>2- If presence of symptoms in 1, verify the BP lying down, sitting and standing at 1-3-5 min intervals<br>3- If orthostatic hypotension documented in 2, refer patient to family physician for health management. Inform patient of risks associated with dizziness and falling.<br><br>**Hypertension**<br>1- If hypertension: re-take BP before the end of the evaluation.<br>2- If hypertension confirmed ≥140/90., refer to general practitioner. |

People with DM1 often show abnormalities of blood pressure, which is most often in the form of hypotension.[6, 87] When compared with the general population, the systolic arterial pressure of people with DM1 is approximately 20 to 30 mm Hg less and the diastolic arterial pressure is also reduced by 10 mm Hg [6]. Even if the mechanisms that contribute to the presence of hypotension in people with DM1 have not yet been elucidated, two principal mechanisms have to date been described. The first mechanism would be the occurrence of reduced muscle tone in the smooth muscles of the vascular walls of the arteries. The second would be the reduction in cardiac output, which can occur in the presence of bradycardia.[34]

**NURSING INTERVENTIONS**

Management is only required when hypotension becomes symptomatic. Refer patient to his general practitioner if symptoms. There is no DM1 specific intervention. Follow national clinical guideline for hyper- or hypotension.

# EVALUATION OF THE GASTROINTESTINAL SYSTEM

It has been observed that smooth muscles of the gastrointestinal system are more affected in DM1 than in any other neuromuscular disease. The symptoms linked to smooth muscle impairment are not only surprisingly common, but can also be the initial clinical characteristic or may dominate the clinical image.[6, 102] Knowing the diverse gastrointestinal manifestations of DM1 can facilitate prompt recognition of the symptoms and efficient health management.[102] Misguidedly, the symptoms are often underestimated or ignored. [103]

## GASTROINTESTINAL SYSTEM

## ELEMENT OF SURVEILLANCE
Anthropometric data

| ANTHROPOMETRIC DATA | | | | |
|---|---|---|---|---|
| **Evaluation** | | | | **Reference/Intervention Criteria** |
| Height<br>Actual weight (lb or kg)<br>Mesured ☐<br>☐<br>Reported by client ☐ | _____<br>_____ lb<br>_____ kg | Weight loss > 5% during the last year?<br>• Is there a reason why you lost weight during the last year? | Y ☐ N ☐<br>Y ☐ N ☐ | *If yes, specify:* _____<br>_____<br>*If no, assess nutritional deficit. If presence of a nutrional deficit, provide information or refer to a nutritionist.* |
| Weight at last appointment (lb or kg) | _____ lb<br>_____ kg | | | _____ |
| | | Weight gain > 5% during the last year?<br>• Is there a reason why you gained weight during the last year? | Y ☐ N ☐<br>Y ☐ N ☐ | *If yes, specify:* _____<br>_____<br>*If no, provide information or refer to a nutritionist.* |
| Waist circumference (cm) | _____ cm | • Man: if > 102 cm, refer to a nutritionist.<br>• Woman: if > 88 cm, refer to a nutritionist. | | _____ |

A study involving 200 DM1 patients shows that 21.0% of people are obese (BMI> 30) and 32.5% are overweight (BMI between 25-30).[104] In another study of 27 people with DM1, 10% were obese (BMI> 30) and 13% had a BMI value in the underweight category.[105] But the BMI is not a good indicator in DM1 as it tends to underestimate the prevalence of obesity due to impaired muscle mass. The measurement of waist circumference can be used in clinical practice as a first step and as a gross indicator in identifying those with excessive accumulation of visceral adipose tissue (VAT).[106] VAT could play a central role in the explanatory mechanisms of the metabolic syndrome.[107] However, due to weakness of abdominal muscles in DM1, it is not clear whether the waist circumference measure is more reliable than the BMI.

In the general population, the probability of hypertension increases by 10% and pre-hypertension increases by 5% with each additional centimeter above the normal waist circumference. Waist circumference greater than 102 centimeters for men and 88 centimeters for women result in a higher risk of diabetes and metabolic syndrome.[106] In DM1, the same observation needs to be validated while taking into consideration the low prevalence of hypertension and diabetes in this population.

Few studies have looked the underweight factor associated with the DM1 population, but it could result from dysphagia.[108]

**NURSING INTERVENTIONS**

There is no DM1 specific intervention. Follow national clinical guideline for weight management.

## GASTROINTESTINAL SYSTEM

ELEMENT OF SURVEILLANCE

Nutrition

| NUTRITION | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| How many meals do you take per day? | _____ | If necessary, provide information about healthy food habits |
| Do you follow a particular diet? | Y ☐ N ☐ | If yes, provide information or pertinent documentation. |
| Assess the quality of the diet (clinical judgment, note quality on a scale from 1 to 5) | _____ | 5 represents a very balanced diet. |
| Do you have difficulty holding utensils?<br>Do you have difficulties preparing meals? | Y ☐ N ☐<br>Y ☐ N ☐ | If yes, refer patient to an occupational therapist for adaptations or home services for meal preparation. |

Nutrition is related to several aspects including transportation to the supermarket, choosing foods, cooking and eating. The quality of the nutrition may depend on several factors including facial weakness, tongue myotonia, oropharyngeal dysphagia, hypersomnolence, cognitive impairment, gastrointestinal dysmotility, and distal muscle weakness and hand myotonia.[109] A study of 29 people with DM1 showed that 62% did not meet the daily-recommended amount of energy intake but the consumption of protein seemed nevertheless adequate. Furthermore, 55% of people with DM1 had a daily consumption of fats above the recommended intake of macronutrients. With regards to intake of macronutrients, a large proportion of people in the study did not meet the daily recommended intake and between 21 and 100% did not consume enough vitamin E and mineral (calcium, magnesium).[105, 110]

The intervention, the nurse must take into account includes all the characteristics of the food environment such as geographical accessibility, availability, cost and quality of food.[111] It is therefore essential to understand the eating behavior of the person. It must be evaluated from an ecological perspective and take into account the individual, social, environmental, organizational and public policies aspects.[111]

## GASTROINTESTINAL SYSTEM

### ELEMENT OF SURVEILLANCE
Oral health

| Evaluation | | Reference/Intervention Criteria |
|---|---|---|
| Dentures?<br>• If yes, age of dentures: | Y ☐ N ☐<br>_____ | _____ |
| How often do you brush your teeth? | _____ | If less than once a day, proceed with education. |
| When was your last visit with the dentist/denturist? | _____ | If the last visit is over a year ago, or if there is any particular problem, refer patient to appropriate specialist. |

Although the prevalence of oral health problems has not yet been documented in DM1, it is generally recognized that frequent oral health problems arise in this population, most notably poor dental hygiene and cavities. When compared with a general population, two studies show that the following factors contribute to a higher rate of cavities in the DM1 population: longer clearance of sugar in the mouth, less secretion of saliva, greater consumption of foods rich in concentrated carbohydrates, increased presence of dental plaque, less efficient coordination of mouth muscles, and difficulty performing oral care due to distal hand weakness, myotonia and reduction in fine motor skills.[112, 113]

**NURSING INTERVENTIONS**

Education concerning oral hygiene should follow national guidelines. However, clinical experience has shown that electrical toothbrush and flossing aids could compensate for lack of hand strength.[114]

## GASTROINTESTINAL SYSTEM

## ELEMENT OF SURVEILLANCE
Abdominal pain

| ABDOMINAL PAIN | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have abdominal pain?<br>• If yes, how often? | Y ☐ N ☐<br>_____ | If yes, and the frequency is greater than once a week and the pain interferes with activities of daily living proceed with nutritional education and refer patient to his general practitioner. |

In a study of 40 people with DM1, it was observed that 25% suffered from abdominal pain.[115] People with DM1 often complain about abdominal pain caused by a dysfunction of the small intestine or by a dysmotility of the colon.[102, 115] The clinical portrait appears in the form of spastic colon or irritable bowel syndrome and is often accompanied by diarrhea or constipation.[102] Gallbladder disease should also be considered in the differential diagnosis of abdominal pain.[6]

**NURSING INTERVENTIONS**

Nursing interventions are focussed on improving transintestinal elimination (see diarrhea and constipation section). There is no DM1-specific intervention. Follow national clinical guideline for irritable bowel management.

## GASTROINTESTINAL SYSTEM

## ELEMENT OF SURVEILLANCE
Constipation/Diarrhea

| CONSTIPATION/DIARRHEA | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you suffer from constipation?<br>• If yes, how often? | Y ☐ N ☐<br>_____ | If the frequency is greater than once a week, proceed with nutritional education and refer patient to his general practitioner. |
| Do you suffer from diarhea?<br>• If yes, how often? | Y ☐ N ☐<br>_____ | If the frequency is greater than once a week, proceed with education of personal hygiene techniques and nutrition and refer patient to his general practitioner. |

Diarrhea and abdominal cramps are often seen (up to 35%) in people with DM1.[102, 115] Constipation and abdominal discomfort can be induced by megacolon, the absence of segmental contractions and loss of peristaltic activity.[116] Intestinal pseudo-obstruction can be the first clinical manifestation of DM1.[117] Intestinal pseudo-obstruction[118], a severe intestinal motility disorder, is provoked by a dysfunction of the myenteric plexus neurons of smooth muscle cells.[117] Mechanisms for diarrhea in DM1 include the mal-absorption of bile acids and the bacterial contamination of the small intestine.[102, 119]

**NURSING INTERVENTIONS**

To treat constipation, a variety of options may be necessary ranging from a simple action to a more complex strategy. It can include increased consumption of dietary fibers, liquids, complementary fibers, emollients (docusate calcium) and osmotic laxatives (lactulose).[102] Irritant laxatives and bowel cleansing are usually avoided, but may be occasionally necessary.

## GASTROINTESTINAL SYSTEM

## ELEMENT OF SURVEILLANCE
Fecal and urinary incontinence

| FECAL AND URINARY INCONTINENCE | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you suffer from fecal incontinence?<br>• If yes, how often? | Y ☐ N ☐<br>_____ | If yes, proceed with education of personal hygiene techniques. If the frequency is more than once per month, refer patient to his general practitioner. |
| Do you have urgent urination?<br>• If yes, does it bother you to the point to consider medication? | Y ☐ N ☐<br>Y ☐ N ☐ | _____<br>If yes, refer patient to his general practitioner. |
| Do you suffer from urinary incontinence?<br>• If yes, how often? | Y ☐ N ☐<br>_____ | If yes, proceed with education of personal hygiene techniques. If the frequency is more than once/month, refer patient to his general practitioner. |

Fecal incontinence, the most handicapping of all gastrointestinal symptoms, is a common characteristic of people with DM1.[102, 115] According to DM1-CIHR project, 14% of people with the adult phenotype of DM1 reported fecal incontinence more than once a month. A study involving 40 people with DM1 revealed that 12,5% suffered from fecal incontinence.[115] Involvement of the anal sphincter is probably responsible for the occurrence of soiling and fecal incontinence.[120]

The urinary system is normally not affected in DM1[6] and urinary symptoms are rarely manifested.[121] The urgency and frequency of micturition and stress urinary incontinence are the main problems. However, according to DM1-CIHR project, 21% of people with DM1 reported suffering from urinary incontinence at least once a month. In DM1, muscles of the urinary pathways and a dysfunction of the pelvic nerve are affected and it has been suggested that they are the mechanisms that cause urinary incontinence.[121]

**NURSING INTERVENTIONS**

Treatment of defecation problems is difficult in DM1.[102] Even though conflicting results have been reported with regards to the effects of procainamide, reference to the general practitioner should be done.[122]

Urinary and fecal incontinence may have repercussions for the person both physically and emotionally. For example, at the physical level, incontinence may indirectly increase the risk of falls and injuries[123] because of the reduced mobility due to muscle weakness. People with a urinary or fecal incontinence problem can be embarrassed, afraid or shy. Also, the incontinence can affect leisure and activities of daily life (social isolation)[124] and the inability to adapt or tolerate the symptoms.[125] Moreover, in clinical practice, it was observed that sub-optimal management of incontinence was associated with individuals reducing their fluid and food intake thereby increasing the risk of dehydration or malnutrition.

## GASTROINTESTINAL SYSTEM

## ELEMENT OF SURVEILLANCE
Dysphagia

| DYSPHAGIA | | | |
|---|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** | |
| During the last week, have you choked or coughed while you were eating or drinking solid or liquid food?<br><br>Swallow test: 90ml (3 oz) of cold water:<br>• Does the patient choke on water during the first minute of the swallowing test? | Y ☐ N ☐<br><br>Y ☐ N ☐ | If yes to the question and if the swallow test is:<br>• **abnormal,** educate about the Heimlich maneuver and refer patient to a nutritionist<br>• **normal**, educate about the Heimlich maneuver<br><br>Provide the Heimlich maneuver pamphlet | If in doubt, complete a dysphagia assessment scale. |

In a study of 40 people with DM1, 45% reported having symptoms of dysphagia.[115] During a radiological study, 20% of the results revealed aspiration with or without symptoms of dysphagia.[126] Another study revealed that 20% presented with dysphagia, 15% presented with coughing while eating and 12.5% needed to clear their throats during swallowing.[115] The nature of swallowing problems in DM1 is complex and investigations have revealed abnormalities in the smooth muscles and in the striated muscles. Oropharyngeal dysphagia and esophageal motility disorders have been proven as the most common source of aspiration pneumonia.[127]

Muscular myopathic weakness of the jaw, mouth, tongue, mandibular region and pharyngeal constrictors is responsible for the slow transition of the bolus to the pharynx. Myotonia of the oropharyngeal muscles is a probable contributing factor of swallowing failure. It has also been suggested that the involvement of the central nervous system could contribute the delayed initiation of deglutition reflex.[127] Alterations have been detected during the pharyngoesophageal phase of deglutition in 12 out of 15 people, of whom six presented with no clinical evidence of dysphagia. Incomplete relaxation of the superior esophageal sphincter and esophageal hypotonia were the most common alteration.[126]

**NURSING INTERVENTIONS**

Coughing during the swallowing test has proven to be a positive predictor of aspiration risk at 84%, and a negative predictor at 78%. If only coughing is evaluated, the positive predictive value is 71% and the negative predictive value is 77%.[128]

A Cochrane report concluded that no study has adequately evaluated treatments for the management of dysphagia in neuromuscular diseases.[129] The principal options of treatment are most often based on populations who suffered from strokes and include dietary adjustment, adoption of safe swallowing techniques, surgical interventions and enteral feeding. No effective treatment has ever been described for dysphagia in DM1, probably reflecting the different underlying mechanisms of dysphagia in DM1.[102]

The strategies that aid the pharyngeal swallowing phase in people with DM1 include[130]:

1) Complete compliance to precautions related to gastroesophageal reflux;
2) Educating friends and family on how to perform the Heimlich maneuver;
3) Nutritional counselling which focuses on the food density (consistency levels: liquid, thick liquid, solid and pasty solid);
4) Using strategies that facilitate clearing of the pharynx, such as:
   - carefully chewing until the alimentary bolus becomes a consistent liquid,
   - doing repeated swallows,
   - taking small bites,
   - alternate between liquid and solid consistency during the meal, and
   - eating meats with sauce;
5) Using strategies that protect the airway when the risk of aspiration is elevated. The posture while eating or drinking can protect the entry to the respiratory tract (the larynx). To protect the respiratory tract, it is recommended to:
   - put head down and chin on the chest, and
   - adopt a sitting position, keeping the body as straight as possible.

# GASTROINTESTINAL SYSTEM

## ELEMENT OF SURVEILLANCE
Gastroparesis

| GASTROPARESIS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you experience nausea?<br>• If yes, how often?<br>Do you have vomiting?<br>• If yes, how often?<br>Do you experience early satiety (feel full quickly)?<br>• If yes, how often? | Y ☐ N ☐<br>_____<br>Y ☐ N ☐<br>_____<br>Y ☐ N ☐<br>_____ | If the answer is yes to any one of the questions and the frequency is more than once per week, refer patient to his general practitioner.<br><br>Yes, why? _____ |

In a study of 40 people with DM1, 15% had vomiting and 12.5% suffered from early satiety.[115] Impaired gastric emptying in people with DM1 is characterized by a longer gastric latency, which corresponds to the time required to reduce the size of solids after first being ingested and up until the evacuation of the solids, and an emptying phase that is one and a half times longer than a control group. Symptomatology was not attributed to a specific phase of gastric emptying.[131] In DM1, gastric symptomatology consists of regurgitation, early satiety, nausea and vomiting, bloating and abdominal pain. Even though these gastric symptoms are less frequent compared to those of the esophagus and pharynx, gastroparesis can be present even in the absence of esophageal and pharyngeal symptoms.[102, 132]

**NURSING INTERVENTIONS**

Questions developed in the DM1-ICP for symptoms of gastroparesis are based on Ronnblom studies.[115, 131]

The neuromuscular nurse could inform the patient's general practitioner of the following indications for medical treatment specific to DM1. Several treatments of gastroparesis in DM1 have been studied, including the administration of metoclopramide which has been found to accelerate gastric emptying by increasing the motility of gastrointestinal movements by an unknown mechanism.[102] The prokinetic agent, cisapride, was removed from the United States and Canada because of potential risk of cardiotoxicity. Globally, the interventions showed limited success.[131]

## GASTROINTESTINAL SYSTEM

### ELEMENT OF SURVEILLANCE
Gastroesophageal reflux

| GASTROESOPHAGEAL REFLUX | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you suffer from heartburn?<br>• If yes, how often? | Y ☐ N ☐<br>_____ | If yes and the frequency is more than once per week, proceed with education on modifications of life habits (e.g. avoid eating before going to bed, drinking alcohol and smoking) and on elevating the head while in bed. If the recommendations are inefficient, refer patient to his general practitioner. |

Gastroesophageal reflux has been the object of few studies. Only one study reported gastroesophageal reflux in three people out of 40.[115]

**NURSE INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guideline for gastroesophageal reflux.

## EVALUATION OF THE MUSCULAR SYSTEM

Muscular impairment is the major feature in DM1. It consists of a progressive weakness from distal to proximal muscles and the presence of myotonia. Muscle weakness is bilateral with a greater weakness in distal muscles (figure 4).[93, 133] First symptoms appear in facial and jaw muscles with temporal and sternomastoid muscle atrophy.[6] Flexor muscles are generally weaker than extensor muscles.[93] The most significant weakness is located in the flexors of the neck and trunk.[93, 133] The most affected muscles are the extensors of the wrist, the long flexor and extensor muscles, the intrinsic muscles of the hand and the dorsiflexors of the foot.[133]

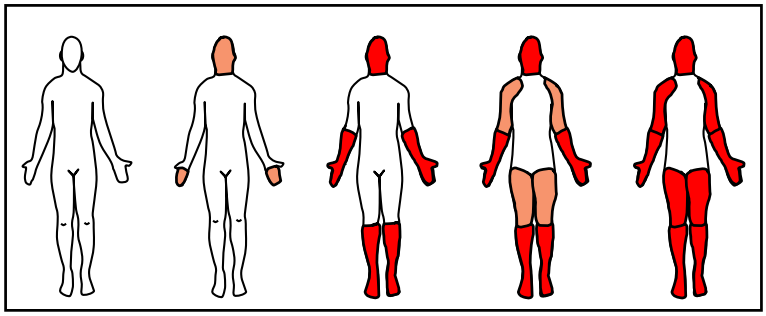

**Figure 1. Progression of the weakness in DM1**[134,135]

**Explanations:**

**Class 1:** No muscular impairment

**Class 2:** Minimal signs Myotonia, jaw and temporal wasting, facial weakness, neck flexor weakness, ptosis, nasal speech, no distal weakness except isolated digit flexor weakness

**Class 3:** Distal weakness No proximal weakness except isolated elbow extensor weakness

**Class 4:** Light to moderate proximal weakness.

**Class 5:** Severe proximal weakness (MRC scale ≤ -3/5).

**Exceptions:** Weakness of the finger flexors happens early on; often no other significant distal weakness is present. People with isolated weakness of finger flexors remain in class 2. Weak brachial triceps often occurs in the absence of proximal weakness; those with this isolated weakness remain in class 3.

At the moment, understanding muscle weakness has been mostly based on histological studies. Studies show that muscle fibers are affected by type I fiber atrophy, which is accompanied by type II fiber hypertrophy and the presence of fat and connective tissue.[136, 137] Decreased muscle strength has been shown in several studies in DM1.[93, 133, 138] A chronic increased of CK is also present[139] and is linked to a rupture of the muscular membrane integrity. It is to be noted that CK levels are included in the evaluation of the muscular system. In one study that was carried out over a two-year period, a minimal but significant decrease of muscle strength was shown in distal and proximal muscles (anterior deltoid, middle deltoid, superior trapezius, quadriceps and anterior tibial).[138] The rate of strength loss is estimated at approximately 1.2% per year.[133] The study also suggested that on a yearly basis, there was faster loss of muscle force in the distal muscle groups than in the proximal muscle groups.

The relationship between the duration of the disease and the loss of muscle strength is not clear. Some studies have shown a high correlation[133], whereas others have refuted this relation [93]. The duration of the disease is calculated from the age of onset of symptoms, which is often difficult to establish probably causing the difference between observations. The loss of muscle strength in the DM1 population is relatively slow .[93] Distal muscular weakness occurs nine years after the onset of the disease (spread between three and 15 years after the appearance of the first symptoms), proximal muscle weakness occurs after 18 years (spread between nine and 27 years after the appearance of the first symptoms) and severe proximal muscle weakness occurs after 27 years (spread between 17 and 37 years after the appearance of the first symptoms).[135] Studies tend to demonstrate that CTG expansion does not explain, or hardly explains, muscle weakness.[137]

***Facial muscles***

The typical facies of people with the adult phenotype of DM1 include immobility of superficial muscles or a reduction of facial expression and weakness in the upper eyelid elevators and orbicular eye muscles, creating a ptosis of the eyelids. Atrophy and weak jaw muscles (temporal and sternomastoid muscles), which result in irregular blockage and weakness during mastication, have also been documented.[6]

***Soft palate muscles***

Weakness of the soft palate muscles can result in speech difficulties, which are typical to the disease, and is associated with the presence of nasal voice. Facio-bulbar weakness can bring about flaccid dysarthria, which translates into myotonia or weakness of the voice.[140, 141]

***Neck muscles***

Weakness and atrophy of the sternomastoid and anterior muscles of the neck can occur at the onset of the disease and difficulty lifting the neck while lying down is often observed.[6] Some studies have shown that half of the subjects had muscle strength inferior to 3/5 during manual muscle testing, which corresponds to a loss of strength about 63% when compared to reference values.[93, 138] The loss of strength of the neck flexors is not linear; a significant loss is observed during the first 20 years and is then stabilized.[133] Clinically, people with DM1 report having difficulty lifting their heads off their pillows and maintaining their neck in an adequate position while driving.[142]

***Hand muscles***

In DM1, weakness in the hand muscles, more particularly in the long flexors, often leads to a grip characterized by the absence of flexion in interphalangeal articulation.[142] The weakness appears quickly in the course of the disease and affects grip strength. In a study, the overall grip strength was 65% weaker than the control group, a result which is similar to previous studies.[93, 138] Men are shown to have a higher overall grip strength than women, which is an observation that is consistent with the general population.[143]

***Elbow extensors***

A weakness of the elbow extensors in DM1[133] may be present; however, the weakness does not interfere with the execution of non-resisted upper limb movement, except when proximal weakness is present.[144]

***Knee extensors***

People with DM1 show significant weakness in the knee extensors when compared with a control group.[138] Knee-extensor weakness is clinically associated with frequent falls.[142]

***Ankle dorsiflexors***

Ankle dorsiflexor weakness is among the first group of muscles which show significant weakness and can bring about foot drop and frequent falls.[133, 142]

**Elements that are present but not included**

People with DM1 often develop nasal voice and speech problems, rendering them sometimes difficult to understand.[145] Flaccid dysarthria was documented in the adult and early adult onset.[140, 145, 146] Even though dysarthria affects the muscular system in DM1, it is not included in DM1-ICP as no efficient intervention has been identified to date. If the DM1 patient brings up the subject, the nurse can provide relevant information or offer support.

## MUSCULAR SYSTEM

### ELEMENT OF SURVEILLANCE
Technical aids: use of a wheelchair

| TECHNICAL AIDS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Mobility: Type of orthosis:<br>Ankle-foot orthosis R ☐ L ☐ _____<br>Cane ☐ _____<br>Quadripod ☐ _____<br>Walker ☐ _____ | Manual wheelchair ☐<br>Motorised wheelchair ☐<br>3 or 4-wheel electric scooter ☐<br>Other ☐ | _____<br>_____<br>_____<br>_____<br>_____ |
| Wheelchair use:<br>• Do you have difficulty using your wheelchair over long distances? | Y ☐ N ☐ | If yes, refer patient to an occupational therapist for a motorised wheelchair. |
| • Year of last repair or maintenance of the wheelchair | _____<br>_____ | _____<br>Refer to appropriate services for annual maintenance.<br>_____ |

According to DM1-CIHR project, between 6% and 17.5% of people with DM1 use a wheelchair.

**NURSE INTERVENTIONS**

Wheelchair and technical aids attribution rules may vary from country to country. There is no DM1-specific intervention. Follow national clinical guideline for wheelchair use.

# MUSCULAR SYSTEM

## ELEMENT OF SURVEILLANCE
Transfers

| TRANSFERS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have difficulty to rise from the toilet?<br><br>Do you have difficulty to get in and out of the bath? | Y ☐ N ☐<br><br>Y ☐ N ☐ | If yes to one of these questions, refer patient to an occupational therapist for technical aids.<br><br>If no, proceed with a visual evaluation in case of doubt. |

According to DM1-CIHR project, 30.5% of people with DM1 require technical aid or human assistance to transfer from the bed to a chair, 29% require technical aid or human assistance to transfer on to the toilet and 49.5% require technical aid or human assistance to transfer into the bathtub**.** The progressive loss of muscle strength can explain the use of technical aids during the course of the disease.

**NURSING INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guideline to refer patients to the appropriate services to obtain assessment and appropriate aids and adaptations.

## MUSCULAR SYSTEM

### ELEMENT OF SURVEILLANCE
Walking

| WALKING | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| During the last month, have you stumble or fall?<br>• If yes, how many times? | Y ☐ N ☐<br>_____ | If more than one fall per month:<br>a) Use the standing test to explore the pertinence of technical aids for walking.<br>b) If the person already uses technical aids for walking other than a wheelchair, determine whether the patient would be ready to receive information regarding the use of a wheelchair. If yes, refer patient to a physiotherapist. |
| **Unipedal balance test (standing test):** Stand on your prefered leg for as long as possible, without using any support (calculate how long the patient is able to stand on one leg). | _____ | If the patient is unable to stand on one leg for 5 seconds, refer patient to a physiotherapist. (this is only an indication of problems but not an absolute rule) |

In a study with a small sample, 58% reported having walking limitations, 31% used technical aids for walking and 6% used a wheelchair.[147] In a study of 13 people with DM1, falls and stumbles were 10 times more frequent than compared to healthy-but-inactive individuals. Walking speed is also lower than the general population.[148] In a larger perspective, mobility is related to being able to travel on short or long distances with or without transportation methods. Restricted mobility and use of different transportation methods are included in the mobility category. In a study of 200 participants, 54.8% reported having no mobility or restricted mobility.[15] The progressive loss of muscle strength of the lower extremities, fatigue, perceived lack of support from family and friends, low level of education and a low revenue explain, in part, the risk associated with having compromised social participation with regards to mobility.[149]

Ankle dorsiflexors are one of the first groups of muscles to show significant weakness, which can cause foot drop and frequent falls.[133, 138, 142] Moreover, knee extensors are weaker when compared to a control group[138] and is clinically associated with frequent falls.[142] During the progression of the disease, the general walking pattern is affected by the distal muscle weakness including the upper leg,

soleus and gastrocnemius muscles.[150, 151] A large study has shown that mobility in general was influenced by several factors including lower extremity strength, fatigue, services and technical aids. Moreover, the access to devices for long distances, such as 4-wheeled mobility scooters, should also be explored considering the high percentage of participants in this study showing poor muscle strength (40%) and fatigue (45%), conditions complicating long-distance activities[149].

**NURSING INTERVENTIONS**

No standardized evaluation is available to assess the need for a physiotherapy referral. According to DM1-CIHR project, the balance test on one foot, one element of the Berg balance scale[152], was selected for the DM1-ICP because it is deemed the best way to discriminate between who used a technical aid or not. Moreover, the number of falls was added to improve the precision of the screening test. The screening test will require further formal validation.

There is no DM1-specific intervention. Follow national clinical guideline to refer patients to the appropriate services to obtain assessment and appropriate aids and adaptations. As well, a home fall prevention checklist should be done with the patients for environmental hazard.

## MUSCULAR SYSTEM

## ELEMENT OF SURVEILLANCE
Myotonia

| MYOTONIA | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Does myotonia disturb you in your daily activities?<br><br>If yes, does it bother you to the point to consider medication? | Y ☐ N ☐<br><br>Y ☐ N ☐ | <br><br>If yes, refer patient to a neurologist. |

Myotonia affects nearly 100% of people with the late or adult phenotype of DM1.[6] Myotonia can be observed when a person is unable to release a muscle after a significant contraction (e.g., after shaking hands with someone). In DM1, myotonia is mainly present in forearm muscles and hand flexors, but sometimes in tongue and jaw muscles.

**NURSING INTERVENTIONS**

The evaluation of the severity of myotonia can be done using more precise instruments; however, they are difficult to administer in clinical context.[153]

The neuromuscular nurse could inform the patient's general practitioner of the following indications for medical treatment specific to DM1. For certain people, myotonia can be quite disturbing and may require medical treatment. The systematic review by the *Cochrane Collaboration* determined that considering the lack of quality data, it was impossible to assess whether medical treatments were safe and efficient in the treatment of myotonia in people with DM1.[154] However, several medications are currently being used such as mexiletine, phenytoin, tocainide and procainamide. Given the possible cardiac complications with these drugs and the potential for malignant arrhythmias, patients should have an ECG before starting treatment.

Nurses can inform the affected person that the presence of myotonia may be reduced by the muscular "warm up" effect. Some vigorous contractions prior to a grasping activity can diminish the phenomenon associated with myotonia.[153] Myotonia can be exacerbated by cold temperature and use of thermal gloves for winter activities is recommended.

## EVALUATION OF THE CENTRAL NERVOUS SYSTEM

Involvement of the CNS occurs in the large majority of patients with DM1[6, 155, 156] particularly when symptoms appear early in life. This can range from a condition of mental retardation (a characteristic often associated with congenital DM1 in which symptoms are manifest from birth), to behavioural changes (e.g. reduced initiative, inactivity, apathetic temperament).[6] Excessive daytime sleepiness is a prominent feature of DM1 and is most often considered as independent from respiratory dysfunction or nocturnal sleep disruption.[157] Moreover, psychopathological disturbances, such as avoidant or paranoid personality traits, are frequent in the adult form of DM1.[158-160] Higher cognitive function disabilities are variably impaired[156, 161-166] but a trend toward reduced frontal lobe performances has often been reported, along with many significant brain changes (e.g. brain atrophy, cell loss, ventricular enlargement, diffuse white matter lesions as well as significant cerebral blood flow reduction in frontotemporal lobe regions), even though correlations are not found in all studies.[6, 167, 168] Previous studies have described the cognitive profile in the classic adult phenotype of DM1, that is generally characterized by mild intellectual disabilities, executive dysfunctions, visuospatial and visuo-constructional disabilities, and learning problems.[155, 169-174] Presently, the potential role of age, disease duration and CTG repeats in the emergence of the cognitive profile in DM1 is not clear. Few recent longitudinal studies on cognition in adult-onset DM1 patients have been conducted. The first study to explore the longitudinal course of cognition in DM1 exhibited no change.[156] Later, evidence of linguistic and executive function abilities deterioration have been found in 34 non-congenital DM1 patients within a 4-year period, correlated with aging but not CTG expansion.[175] The authors interpreted these results as the development of an early fronto-temporal dementia cognitive profile with aging. Others found, after a 7-year follow-up, a selective progressive impairment only in the attentional sphere in 14 patients with DM1, without any correlation to the level of muscle weakness.[176] It is still a matter of debate whether this cognitive profile is a stable feature of the disease or a progressive manifestation of a cognitive decline.

## CENTRAL NERVOUS SYSTEM

## ELEMENT OF SURVEILLANCE
Excessive daytime sleepiness

| EXCESSIVE DAYTIME SLEEPINESS | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Describe your sleep habits (bedtime, the time when you wake up, frequency, schedule and for how long you take naps). | | If the sleepiness interferes with nutrition, leisure or other daily activities, proceed with counseling on behavioural changes and education on how to keep a sleep diary before referral to a neurologist. |
| 1- Do you take naps during the day? | Y ☐ N ☐ | |
| 2- If yes, does it disturb your daily activities? | Y ☐ N ☐ | If yes to questions 2 and 3, refer patient to a neurologist to evaluate the pertinence of a nocturnal polysomnograph and specific treatment. |
| Medication : | | |
| 3- If no medication is taken, does your sleepiness bother you to the point to consider medication? | Y ☐ N ☐ | |
| 4- If taking psychostimulant (e.g., Ritalin, Dexedrine), is it efficient? | Y ☐ N ☐ | If not or hardly efficient, discuss with a neurologist. |

Excessive daytime sleepiness (EDS) has long been associated with DM1.[177] It is the most common complaint and the non-muscular symptom that is most often described.[6, 34, 103, 178] EDS can be one of the first clinical manifestations of the disease as many people complain of EDS years prior to receiving a diagnosis of DM1. EDS was observed in nearly 80% of people with DM1.[103, 179] It was determined that, using the Epworth Sleepiness Scale (ESS)[180] and the Daytime Sleepiness Scale (DSS)[181], approximately 30% of people with DM1 reported daytime sleepiness.[182] Studies suggest that EDS is the result of a primary CNS manifestation[34, 183, 184] and is usually independent of respiratory functions and nocturnal sleep perturbations.[172, 179, 181, 185] Precise etiology remains uncertain and EDS can be linked to fragmented sleep, central and obstructive sleep apneas, hypercapnia and irregularities in paradoxical sleep.[78, 177, 179, 183, 186-188]

**NURSING INTERVENTIONS**

The use of standardized questionnaires to evaluate EDS in DM1 is outside the scope of DM1-ICP. The main objective is rather to identify if EDS is a problem that significantly bothers the person and to assess whether that person desires a referral for an evaluation and specific treatment.

A description of sleep habits can be useful in identifying factors that can interfere with healthy sleep hygiene and evaluates the impact of EDS on daily activities. Moreover, attention should be targeted towards other factors that are not related to DM1 such as alcohol and caffeine consumption, and the secondary effects of some medication, all of which could be modified.[189] Some people with DM1 are able to deal with EDS by taking naps at times and are therefore reluctant to receive clinical evaluation and treatment.

## CENTRAL NERVOUS SYSTEM

## ELEMENT OF SURVEILLANCE
Fatigue

| FATIGUE | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you feel tired every day? | Y ☐ N ☐ | *Specify* _____ |
| Does fatigue prevent you from fulfilling some tasks and responsibilities? | Y ☐ N ☐ | |
| If yes, is it bother you to the point to consider medication? | Y ☐ N ☐ | If yes, refer to a neurologist or to his general practitioner_____ |

Fatigue is more frequent in DM1 than in any other neuromuscular disease. Like EDS, fatigue can be a reason for a first consultation even with people with mild muscular impairment.[6, 190] Significant overlapping between fatigue and EDS exists, whereby the level of fatigue is higher in people with EDS and the level of daytime sleepiness is higher in people with excessive fatigue. The proportion of people with DM1 with an excessive or pathological level of fatigue reaches 62.5% on the Fatigue Severity Scale.[182]

Fatigue is defined as an oppressing feeling of tiredness, a lack of energy and a feeling of exhaustion that must not be confused with weakness.[191] It is not easy to describe the physiology of fatigue in DM1 because of the close ties that exist between fatigue and other associated conditions such as EDS, apathy and depression. In DM1, the fact that some symptoms like apathy, adynamy and lack of motivation are improved with the administration of antidepressants suggests that the behavioral perturbations could be attributed to chronic depression instead of the DM1 disease itself[192], however there is no consensus about this hypothesis. Furthermore, interventions specifically suggested to compensate the motivational aspects in DM1 include the treatment of all depressive episodes.[142] A study suggested that fatigue in DM1 can occur despite normal thyroid functioning.[193]

Like in EDS, the main objective is to identify whether fatigue is a problem that significantly affects the person and to assess the person's desire of being referred for evaluation for specific treatment. The same procedure as with EDS is proposed. Attention should be focussed on other factors that are not related to DM1 such as alcohol and caffeine consumption and the secondary effects of other medications, all of which could be modified.[189]

There is no DM1-specific intervention. Follow national clinical guideline for fatigue.

## CENTRAL NERVOUS SYSTEM

## ELEMENT OF SURVEILLANCE
Depression

| DEPRESSION | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| *Depressive mood*: During the last month, have you often been bothered by feeling down, depressed or hopeless? | Y ☐ N ☐ | If yes to any one of the questions, refer patient to available resources and to his general practitioner AND: |
| *Anhedonia*: During the last month, have you often been bothered by little interest or pleasure in doing anything? | Y ☐ N ☐ | • Evaluate the presence of suicidal thoughts in order to assess imminent risk of suicide:<br>❖ Do you think about suicide or about killing yourself? ☐ N ☐<br>If yes, rate imminent risk of suicide<br>Low ☐ Level _____<br>Medium ☐ Level _____<br>High ☐ Level _____<br><br>Refer to the patient to his general practitioner with more or less emergency depending on the presence of suicidal thoughts.<br><br>Depending on the availability of services offered at the neuromuscular clinic, refer patient to psychologist/neurologist to initiate the evaluation of symptoms like fatigue, agitation, a feeling of guilt, concentration difficulties and changes in sleep habits or appetite with the purpose of confirming the diagnostic impression of depression. |

High scores have been observed in people with DM1 on various clinical scales that measure the intensity of depressive symptoms, in a proportion that sometimes reaches up to 50% of the study group.[161, 192, 194] However, when more restrictive criteria are applied, approximately 17% of people show clinically significant depressive symptoms.[195] Nearly 27% of people with adult or mild phenotype have a positive risk to have a diagnosis of a psychiatric disorder including clinical depression[196], as evaluated by the *Symptom Checklist-90-R*.[197]

For some, depressive symptoms observed in DM1 would result from a genetically determined alteration of the neurotransmission of the central aminergic system[192, 198], whereas for others, the symptomatology would be secondary to the progressive and debilitating nature of DM1.[199] However, it seems that few people present symptoms that meet the criteria associated with an episode of major

depression as indicated in DSM-III-R[200, 201], suggesting that depressive symptoms seem more common than typical depressive episodes[199]. Several questionnaires have been used with various study groups of people with DM1 to measure symptoms of depression, which can, in part, explain the observed differences in the rate of depression. However, at follow-up of people with chronic disease, special attention should be paid to depressive symptoms and the influence that such symptoms may have on the physical and psychological health of the patients.[200] Screening for depression must focus on the identification of one of the following symptoms, either: (1) depressive mood; or (2) loss of interest or pleasure (anhedonia). Symptoms of fatigue, somnolence, apathy or adynamy should be excluded because they may constitute specific symptoms of DM1 and can be confused with depressive reactions.

**NURSING INTERVENTIONS**

From the perspective of general medical follow-up, it is recognized that the integration of systematic screening for depression reduces the persistence of depressive symptoms.[202, 203] It was established that a two-question questionnaire was just as effective as more-detailed screening instruments used to identify probable cases of major depression.[202, 204] The questions of the DM1-ICP were taken from the *Primary Care Evaluation of Mental Disorders Procedure* (PRIME-MD), a screening questionnaire of 27 items that was created to facilitate the diagnosis of some psychiatric conditions during primary care.[202, 203, 205]

The test is considered positive if "yes" is answered to any of the questions. A negative response to both questions indicates that depression is highly improbable. To improve clinical sensitivity, a question on suicidal thoughts was added if the person answers yes to both questions. Estimation of suicidal urgency should be documented using the HWW formula (How? Where? When?).

There is no DM1-specific intervention. Follow national clinical guidelines for depression and suicide prevention.

## CENTRAL NERVOUS SYSTEM

### ELEMENT OF SURVEILLANCE

Sleep apnea / Pain

Sleep apnea and pain are two elements of surveillance that are common in DM1, but there is no consensus on screening procedures and reference/intervention criteria. The elements are always part of ongoing discussions. New modules will be produced in 2014 on these topics.

# EVALUATION OF THE REPRODUCTIVE SYSTEM

Myotonic dystrophy influences the reproductive system in different ways depending of the gender. In women, there may be gynecological problems and there is an increased obstetrical risk. In men, erectile dysfunction occurs. Although the safety of sexual activity is not related *per se* with the DM1 conditions, promotion of safe sexual activity in men and women should be done ensuring that the patient also understands the disease transmission risk to offspring.

## REPRODUCTIVE SYSTEM

## ELEMENT OF SURVEILLANCE

Sexual activity

| SEXUAL ACTIVITY | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Are you sexually active?<br>• If active, do you use protection against STI during sexual relations?<br>• If sexually active, what types of contraceptions do you use?<br>• If you do not have a definitive type of contraception and you want a child, are you conscious of the obstetric risks in DM1? | Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐ | If no or the use of methods is risky, proceed with education on effective contraceptive methods and risk of transmission of sexual diseases.<br>If no, provide information about obstetric risks. |

The infection rate and the rate of protection against STI have never been documented in DM1.

**NURSING INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guidelines for promoting safe sexual activity.

## REPRODUCTIVE SYSTEM

### ELEMENT OF SURVEILLANCE
Male sexuality

| MALE SEXUALITY | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| When you are sexually stimulated, do you have erectile problems? | Y ☐ N ☐ | |
| If yes, does it disturb you? | Y ☐ N ☐ | |
| If yes, is it bother you to the point to consider medication? | Y ☐ N ☐ | If yes, refer patient to his general practitioner. |

According to DM1-CIHR project database, 36.7% of men with DM1 reported mild to severe erectile dysfunction (ED). The results can be an underestimation as 46% of the study group (n = 200) affirmed to have had no sexual relation during the last six months and were therefore not evaluated for ED. In another study, 24.1% reported being impotent, whereas 34.4% reported having erections but no sexual relations. In the most severely affected group, all men reported having no sexual relations.[206] A large investigation about sexuality and ED in the adult Canadian population showed a prevalence of ED affecting 27% of sexually active men.[207] Moreover, a reduction of spermatozoids was also reported in DM1, but there was no significant reduction of fertility.[6]

Impotence can be caused by hypogonadism (see hypogonadism in the Endocrine System section) and by a degeneration of smooth muscle cells in the penis.

**NURSING INTERVENTIONS**

The DM1-ICP is based on a question of the *Sexual Health Inventory for Men* (SHIM)[208], a validation of the International Index of *Erectile Function version.*[208, 209]

There is no DM1-specific intervention. Follow national clinical guidelines for male sexuality.

## REPRODUCTIVE SYSTEM

## ELEMENT OF SURVEILLANCE
Female sexuality

| Female Sexuality | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Have you undergone a hysterectomy?<br>• If no, do you suffer from menstrual problems?<br>• If yes, does it disturb you in your daily activities? | Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐ | If yes, refer patient to his general practitioner. |

***Gynecological problems***

Harper (2001)[6] studied 44 adult women with DM1 and 25 non-affected women but who were related to or the spouse from the same families. This study showed that those with DM1 had a higher frequency of excessively painful menstruations (57.5% compared to 27.3%) and excessively irregular menstruations (35% compared to 23.8%) than those in the control group. Regardless of menstrual problems, the possibility of gonad abnormalities in women with DM1 has received little attention.[6]

***Obstetric risks***

The risk of obstetric complications are increased in women with DM1 as this disease does not only affect voluntary muscles but also affects smooth muscles, namely those of the genitourinary system.[34] Women who developed DM1 early are more likely to have complicated pregnancies than those who developed the disease later on in life.[210]

The spontaneous abortion rate in women with DM1 is two or three times higher than the 10% rate of the control group.[6] A study with 31 women with adult phenotype of DM1 focused on 66 children during 64 gestations and revealed a more elevated frequency of obstetric complications for the DM1 population than for the control group.[211] The complications were also validated by a case review, which included a total of 93 described gestations. These complications are: ectopic pregnancy, placenta previa, polyhydramnios, premature birth (<36 weeks), assisted delivery, caesarean, delivery and perinatal mortality (between the 28$^{th}$ week of gestation and the 28$^{th}$ post natal day).[212] Spontaneous abortions in DM1 is probably caused by abnormal activity of the uterus muscle or by an abnormal level of hormones originating from the gonads.[6] The causes of an ectopic pregnancy have not yet been documented, but it could be related to motility alteration of the fallopian tubes.[212] Bleeding observed during the second

and third trimesters are often associated with an abnormal attachment of the placenta. Indeed, placenta previa in women with DM1 can be the result of uterine dysfunction or perturbations related to the development of the endometrium.

Polyhydramnios (excess of amniotic fluid in the amniotic bag) can be detected between the 31st and the 35th week of gestation, but can also be observed as soon as the 24th week. Polyhydramnios happens almost exclusively during the gestation of a fetus with congenital myotonic dystrophy and is probably due to deglutition problems of the fetus. It is important to note that in pregnancies with polyhydramnios, the premature contractions and severe neonatal distress are elements that require constant surveillance. Other causes can explain premature contractions such as placenta previa and intra-uterine hemorrhages. Complications related to the urinary system have received little attention.[212]

Other complications were observed such as an increased risk of severe urinary infection (13%), vaginal bleeding during the second and third trimesters, prolonged contractions during the first and second phases of delivery and postpartum haemorrhage.[212] Abnormalities have been noted during the three phases of labour. Prolonged labour during the first phase could be related to uterine dysfunction, whereas prolonged labour during the second phase could be explained by maternal weakness and lack of personal willingness. Prolonged labour can cause foetal distress and justify assisted delivery interventions (forceps, suction cup or caesarean).[212] Postpartum haemorrhage during the third phase of labour is caused by inadequate uterine contractions or abnormal attachment of the placenta. Perinatal mortality (pre and neonatal mortality) is mainly attributed to a foetus with congenital DM1 and due to complications during pregnancy.

After childbirth, most women with DM1 return to the previous level of functioning within a short period of time and the course of the disease is not affected by any adverse effect of pregnancy.[6, 211] Anaesthesia risks are present if analgesia or anaesthesia is administered during delivery.

**NURSING INTERVENTIONS**

Counselling on obstetric risks must be done with all women with DM1 who wish to have a baby. The high obstetric risks in DM1 requires constant surveillance in addition to intensive obstetric and perinatal care.[211] In case of pregnancy in women with DM1, the nurse must:

- Refer for genetic counseling to offer:
    - Prenatal genetic analysis and
    - Genetic counseling (see Genetics section);
- Assure adequate medical health management; and
- Assure that the client is known by psychosocial services and that she is registered in a nutritional program for pregnant women.

## RISK FACTORS

### ELEMENT OF SURVEILLANCE
Genetics

Genetic counseling is definitely one of the most important aspects of the clinical management of people with DM1 and their families. Genetic counseling in DM1 is complex and may be extremely difficult for multiples reasons, such as:[34]

- Variability of clinical manifestations (severity and age of onset);
- Perceptions regarding progression and severity of the disease can differ due to this variability;
- Reluctance of elderly people to seek medical advice; and
- Uncommon clinical situations in which the recognition of a genetic disease in the family is perpetuated by the birth of a severely affected child even though other family members may already be clinically affected.

## RISK FACTORS

## ELEMENT OF SURVEILLANCE:
Genetics
Family history

| FAMILY HISTORY | |
|---|---|
| **Evaluation** | **Reference/Intervention Criteria** |
| Gather information about family history particularly about first degree relatives.<br>If family tree chart exist, verify the chart prior to patient's visit and complete the tree with the new information after patient's visit. | |

**NURSE INTERVENTIONS**

Completing the family history is the first step before discussing the transmission risk of DM1. The family tree chart is an extremely useful tool for:

- Gathering genetic information about family members;
- Quickly understanding the family structure including multiple marriages, complex consanguinity and kinship;
- Identifying carriers and non-carriers of the DM1 gene; and
- Evaluating the genetic risk of healthy parents.

Considering that outlining the family tree chart may divulge sensitive family issues, the patient may be confronted with delicate situations (re-activation of familial difficulties or personal events not spoken of…). The nurse must be prepared to provide psychological support when needed. With each visit, the nurse should review the family tree chart and indicate any changes that have taken place since the last visit such as births, deaths or even a new genetic status of family members.

## RISK FACTORS

### ELEMENT OF SURVEILLANCE:

Genetics
Risks for children and family planning if between 18 and 45
Risks for other family members

| RISKS FOR CHILDREN AND FAMILY PLANNING IF BETWEEN 18 AND 45 | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have children?<br>If yes, how many? | Y ☐ N ☐<br>_____ | If one or more children are younger than 18, verify the parents' knowledge with regards to heredity of the disease. |
| If you have children older than 18, have they received genetic testing/screening? | Y ☐ N ☐ | If no, inform the patient of the following:<br>a) potential risks;<br>b) possible genetic counselling referral; and<br>c) his own role as an agent of information for other family members. |
| If you have children younger than 18, do they have any symptoms? | Y ☐ N ☐ | _____ |
| Do you want to have children or more children? | Y ☐ N ☐ | If no, verify contraception.<br>If yes, verify patient's knowledge with regards to heredity, obstetric risks and refer patient to genetic counselling when appropriate. |
| **RISKS FOR OTHER FAMILY MEMBERS** | | |
| **Evaluation** | | **Reference/Intervention Criteria** |
| How many living brothers and sisters do you have? | _____ | _____ |
| If one or more are alive, did they receive genetic counselling? | Y ☐ N ☐ | If no, inform the patient regarding:<br>• his role as an information agent for other family members;<br>• potential risks for other family members; and<br>• possible referral to genetic counselling |

The autosomal dominant transmission, the incomplete penetrance and the variability of age of onset explain the high frequency of parents being unknowingly affected in families where DM1 has been identified.

The risk of transmission of DM1 corresponds to the probability of having received or of transmitting the mutated gene. Considering that DM1 is an autosomal dominant disease, only one of

the two parents needs to be a carrier of the genetic abnormality. Estimation of risk if the other parent does not have the genetic abnormality is the following[6]:

**Estimation of risk**

With each pregnancy:

- A 50% risk (1 out of 2) that the child receives the genetic abnormality and that he/she is affected
- A 50% risk (1 out of 2) that the child does not receive the genetic abnormality.

**Origin of the disease**

DM1 is caused by an abnormal amplification of a CTG trinucleotide repeat on the chromosome 19q 13.3.[9, 26, 213] The number of triplet repeats in an unaffected individual is between 5 and 35. In DM1, between 50 and several thousand CTG repeats are observed.[27]

**The phenotypes**

The clinical classification of DM1 is divided into four clinical phenotypes. The phenotypes are determined by the age of onset and the number of CTG repeats.

**Table 1. Clinical classification of myotonic dystrophy [6]**

| Type | Age of Onset | Principal Characteristics | CTG Repeats |
|---|---|---|---|
| Mild phenotype | >40 | Cataracts, weakness and myotonia are often present | Between 50 and 150 repeats |
| Adult phenotype | 10-40 | Atrophy of facial and sternomastoid muscles, ptosis of the eyelids, excessive daytime sleepiness, chronic fatigue, muscular weakness, and myotonia | Between 100 and 1000 repeats |
| Childhood phenotype | 1- 10 | Variable degrees of developmental delay, variable hypotonia, myotonia develops after childhood | Between 500 and 2000 repeats |
| Congenital phenotype | In utero and at birth | Hypotonia, respiratory failure, facial diplegia, difficulty sucking, and congenital contractures | |

**Correlation between the phenotype and the genotype**

The correlation between the phenotype and the genotype is limited. Most individuals with an expansion of 50-80 repeats are susceptible to being and staying stable in terms of the progression of the disease and could have cataracts as the only significant manifestation.[34] Some individuals with less than 100 repeats show no serious neuromuscular problems.[214] At the other end of the spectrum,

individuals with more than 1,000 repeats are generally severely affected and can have the childhood phenotype, or, if transmitted by the mother, the congenital phenotype.[215] Between the two ends of the spectrum, there is a small correlation between the phenotype and the genotype and the prognostic estimation is probably better if it is based on a careful longitudinal clinical evaluation than on the molecular status of the individual. Therefore, particular caution must be put when trying to estimate the prognosis of the severity of the disease based on molecular results only.

**Incomplete penetrance of the gene**

Penetrance corresponds to the proportion of carriers of the genetic abnormality who actually develop the disease. In DM1, the penetrance is incomplete, meaning that a certain number of individuals (estimated between 5 and 10%) are carriers of the mutation (in general between 50 and 150 CTG repeats) but will not show any signs of the disease during their whole life.[6]

**Anticipation between generations**

Anticipation is the tendency in certain genetic disorders --- like myotonic dystrophy --- for individuals in successive generations to present with symptoms at an earlier age and/or with more severe manifestations; often observed in disorders resulting from the expression of a trinucleotide repeat mutation that tends to increase in size and have a more significant effect when passed from one generation to the next..[6, 10, 216] Usually, children inherit more CTG repeats that those of the parent carrier.[34]

**Effects of parental origins**

Although anticipation is observed both in paternal and maternal transmission, a decrease in the CTG repeat size during transmission from parents to child can also occur in about 6.4% of transmissions, most frequently during paternal transmissions.[217] The congenital form is almost always transmitted by the mother.

**Genetic counselling**

Most genetic-related questions are usually discussed at the moment of the diagnosis, during periodical visits at the clinic or again during home-care visits done by nurses. The previously mentioned opportunities are excellent for assuring that all the aspects related to genetic counselling are properly discussed. The following genetic aspects concerning DM1 must be well understood by those affected and must be periodically re-evaluated[34]:

- autosomal dominant heredity;
- the origin of the disease: unstable mutation (expansion of CTG repeats on chromosome 19);
- extreme variation of severity, age of onset and evolution of the disease as per the different phenotypes;
- the incomplete penetrance of the gene (approximately 90% at adult life);
- anticipation ;
- the effects of parental origins: the congenital form is almost always transmitted by the mother;
- the limited relationship between disease manifestations and the size of the repetition ;
- prenatal diagnosis.

The nurse should also inform patient of his personal role as an agent of information for other family members. As it is often impossible to have direct contact with all at-risk parents of DM1 patients, try to convince those who have already been tested (positive or negative) to inform family members of the genetic aspects of DM1 and the clinical services available for investigation and follow-up.

**Family planning**

Questions pertaining to family planning are only appropriate if the person is of reproductive age. Referring to genetic counselling is indicated for any person with DM1 who desires having children, regardless of gender. Different aspects should be discussed by the genetic counsellor including the composition of the current family, future family planning, birth control, prenatal diagnosis and alternative choices regarding family planning.

**Predictive DNA testing**

The estimated risk of hereditary transmission and the fact that other family members of people with DM1 may be affected have been well summarized by Harper (2001).[6] Predictive DNA testing should be explained and offered to all adults who are at risk. Incidentally, directives published by the International Myotonic Dystrophy Consortium propose that individuals who have predictive testing should always receive pre-test genetic counseling by a qualified counselor.[27] Predictive testing protocols vary slightly per clinical setting.[218, 219] A protocol, which consists of specific elements, must be followed to reduce to a minimum the negative impact perceived by many asymptomatic carriers of the gene regardless of the pre- and post- test systematic counseling. The protocol consists of:[219]

- careful pre-test counseling;
- pre-test clinical evaluation; and
- post-test psychological support and follow-up for identified carriers.

The pre-test predictive counselling must include the following elements:[220]

- verify the reason behind the request for predictive testing at this point in time and correct if there is a false concern regarding symptoms that are not associated with DM1;
- verify the familiarity of the phenotype and the ability to identify the people affected by DM1 within the extended family and to name the signs of the disease;
- provide additional information on symptomatology (age of onset, first symptoms, weaknesses, others) regarding DM1;
- inform of the mode of transmission.

**Prenatal diagnosis**

- Prenatal diagnosis consists of searching for the genetic abnormality (abnormal number of CTG repeats) during pregnancy. Between the $10^{th}$ and the $11^{th}$ week of gestation, the prenatal diagnosis is done by choriocentesis (chorion biopsy) and between the $15^{th}$ and the $19^{th}$ week by amniocentesis. These invasive investigations aim at sampling cells that originate from either the chorionic villi (little projections that develop on the embryonic envelope that constitute as the future placenta) or the amniotic fluid. The procedures permit to collect a sufficient quantity of DNA to establish a diagnosis.

## RISK FACTORS

## ELEMENT OF SURVEILLANCE
Smoking

| SMOKING | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you smoke?<br><br>If yes:<br>• Number of cigarette smoked<br><br>• Any previous attempts to quit smoking tobacco? | Y ☐ N ☐<br>Cigarettes ☐<br>Other ☐<br><br>_____number/day<br><br>Y ☐ N ☐<br>Number of previous attempts: _____ | |
| Does someone in your home smoke? | Y ☐ N ☐ | If yes, proceed with education about the effects of second-hand smoke. Provide documentation on second-hand smoke. |

A rate of 23.6% of regular tobacco use and a rate of 6.5% of occasional tobacco use was observed [104]. These rates are comparable with regional prevalence (29.9% of regular and occasional smokers)[221], but superior to the national prevalence (21%).[222] It was equally noted that people with the adult phenotype had a higher rate of smoking than those with the mild phenotype of DM1.

Pneumonia and chronic respiratory failure followed by cardiovascular disease[13] constitute the most important causes of death in DM1 and can be exacerbated by tobacco use.

**NURSING INTERVENTIONS**

All people with DM1 should be questioned regarding use of tobacco products. Information obtained allows, on the one hand, to adapt the intervention in relation to the person's interest in smoking cessation and, on the other hand, to assure a personalized referral to a Quit Smoking Centre if the person desires assistance.

There is no DM1-specific intervention. Follow national clinical guidelines for smoking cessation.

# RISK FACTORS

## ELEMENT OF SURVEILLANCE
Consumption of alcohol and drugs

As reported by one study, the prevalence of drug use among the DM1 population is 7% and the prevalence of excessive alcohol consumption is 3.5%.[104] However, this prevalence may be underestimated because of the sensitive nature of the information. Indeed, in clinical practice, many patients use drugs to manage their anxiety or insomnia disorder. As the socio-economic conditions of people with DM1 are often compromised and the consequences of excessive alcohol and drug consumption are significant, it may be relevant to assess the consumption of these substances by people with DM1.

### NURSING INTERVENTIONS

There is no DM1-specific intervention. Follow national clinical guidelines for alcohol and drugs consumption.

# SOCIAL PARTICIPATION

Social participation consists of daily activities and social roles that can be divided into 12 broad categories. For the purpose of the DM1-ICP, some activities were addressed in the evaluation of the organic systems and other activities will be addressed in the present section. It is important to highlight that the level of participation of those with the adult phenotype was found to be inferior to the level of participation of an aged population living in the community and that, regardless of the fact that the age of those with DM1 was inferior to that of the control group.[15] Furthermore, between 45% and 61% of participants reported needing help or being unable to accomplish daily activities in the four most disrupted participation categories: mobility, housing, employment and recreation[15]. The majority of the participants were satisfied with their participation in the different spheres of life, except concerning mobility, employment and recreation. Similar results were shown in another study.[223] Satisfaction was strongly correlated with the level of accomplishment of life habits, reinforcing the necessity of focusing on establishing effective interventions in the most disrupted spheres.

Of the personal factors, a low level of education, reduced muscle strength and presence of fatigue are associated with an elevated risk of resorting to help or not accomplishing life habits related to mobility, housing, recreation and employment. Moreover, fatigue is an important factor to consider.[149]

Consideration of environmental factors is also important in ensuring a better understanding of the social participation of people with DM1. Of the environmental factors, the perception that a lack of family and friend support can be an obstacle associated with a higher risk of seeing one's social participation being compromised. The lack of governmental services is associated to several categories of participation as well. Fifty-one per cent of participants perceived that public and governmental services, as currently offered and organized, were an obstacle in their participation in their most disrupted spheres of life.[149] There is therefore great potential to take action to help improve the participation of people with DM1. People with DM1 and their close family and friends are the ones who can provide the most information allowing for a better understanding of the necessary services.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Personal hygiene

| PERSONAL HYGIENE | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have difficulty taking a bath or shower? | Y ☐ N ☐ | If yes, document the cause(s) and proceed with a referal to an occupational therapist or bathing help services. |
| How often do you take a bath or shower? | _____ | If less than once per week, proceed with information or provide a referal to community services. |
| Do you have difficulty getting dressed? | Y ☐ N ☐ | If yes, proceed with education or refer patient to an occupational therapist |

According to DM1-CIHR project, people with DM1 reported having difficulties to take a bath (42%), to use the toilet (22%) and to get dressed (15%). Difficulties encountered by people with DM1 can be related to upper extremity (loss of grip strength, myotonia), lower extremity (loss of strength) functioning or both.

**NURSING INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guidelines for personal hygiene.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Housing: Housework

| HOUSEWORK | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have difficulties with the maintenance of your home ? | Y ☐ N ☐ | |
| Visual inspection if at home:<br>Acceptable (clean)<br>Poorly maintained but acceptable<br>Unhygienic<br>Unsanitary | <br>☐<br>☐<br>☐<br>☐ | If presence of difficulties or if housing is unhygienic or unsanitary, proceed with referal to home careservices. |

A study addressing the participation in housework (cleaning, washing, making food and shopping) showed that 32.6% of people with DM1 were unable to accomplish some activities and 25.8% experienced difficulties accomplishing these activities.[223] According to DM1-CIHR project, 63.5% of the 200 participants reported encountering severe barriers when trying to accomplish heavy housework and 43.5% encountered severe barriers while performing housekeeping of their home.[15] The main factors where excessive fatigue and lower extremity strength.[149]

**NURSING INTERVENTIONS**

A residence is defined as unsanitary if it is considered improper for habitation and constitutes a risk (e.g., contamination, fungi infestation) for the health or for the security of the occupants.

There is no DM1-specific intervention. Follow national clinical guideline to refer patients to the appropriate services to obtain assessment and appropriate services.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Adaptations

No data is available but adaptations the most frequently used based on clinical practice are grab bars in the bath and next to the toilet and an elevated toilet seat.

**NURSING INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guideline to refer patients to the appropriate services to obtain assessment and appropriate aids and adaptations.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Transportation

| TRANSPORTATION | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| **Car driving and transportation:**<br>• Do you have a driver's license?<br>• Do you currently drive?<br>• Do you have a disabled parking permit?<br>Do you use adapted transport? | Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐<br>Y ☐ N ☐ | Refer patient to his general practitioner or neurologist if the abilities related to car driving seemed compromised (e.g., daytime sleepiness, muscle weakness, myotonia, reduced vision or cognitive functioning).<br>_____<br>_____ |
| **Running errands:**<br>Do you have difficulty running errands? | Y ☐ N ☐ | If yes, refer patient to community services . |

According to DM1-CIHR project, 58.5% of the participants still driving a car.

**NURSING INTERVENTIONS**

Several factors must be taken into consideration for the screening of one's driving ability[224]:

- Presence of excessive daytime sleepiness;
- Weak grip strength;
- Myotonia;
- Less than 20/50 visual acuity as per the Snellen chart;
- Presence of arrhythmias;
- Consumption of alcohol or drugs; and
- Affected upper cognitive functions.

The assessment for disabled parking permit and adapted transport should be based on available local eligibility criteria.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Employment

| EMPLOYMENT | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Are you currently working?<br>If no, have you ever worker? | Y ☐ N ☐<br>Y ☐ N ☐ | _____<br>_____ |
| • If yes, do you have difficulty completing tasks related to your work? | Y ☐ N ☐ | If yes, refer patient to local employment resources. |
| • If no, have you made a request for disability pension? | Y ☐ N ☐ | If no, proceed to information and guidance. |
| • If no, would you like to have employment? | Y ☐ N ☐ | If yes, refer to available services. If no, provide information on how to obtain disability pension. |

The rate of employment of people with DM1 is inferior to the general population. In a recent study, 20% of the participants were employed, 66% were once employed and 14% had never been employed.[19] Moreover, 44.5% reported that their participation in their employment was severely restricted and was associated with a high level of dissatisfaction.[15] In previous studies, the percentage of participation in the workforce varied between 12.4% and 31%.[16, 225] People with DM1 have the lowest rate of employment compared to other neuromuscular diseases.[225] A significant difference in the employment rate is observed between men and women within a DM1 population.[226, 227] A study completed in 1983 showed that one out of two women had never worked whereas for men, only one out of five had never worked.[227] In another study, no difference between the genders in terms of employment rate have been reported.[225]

Factors influencing the ability to work in the general population are generally gender, age and education.[226] For people affected by a neuromuscular disease, the factors related to the significant possibility of being unable to maintain employment are: gender (female), age (being older than 30), low level of education, reduced mobility, reduced communication and cognitive capacities, limited movement of upper extremities, and requiring help to complete other life habits.[226] An increased chance of maintaining employment was related to how working conditions were adapted to the needs of the person. The adaptations included a reduced work speed, reduced hours and latitude with regards to the

organization of the work.[226] For people with DM1, lack of access to technology, weakness of lower extremities, fatigue and pain were predictive factors of compromised participation in employment activities.[149]

**NURSING INTERVENTIONS**

Employment services are often poorly informed of the realities of people with DM1. Nurses must make a personal referral to employment services and assure that rehabilitation services are included in the process.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Leisure

| LEISURE | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you have any activities or hobbies?<br>• If yes, how often do you do them?<br>_____<br>_____<br>_____ | Y ☐ N ☐<br>_____ | If no, discuss the possible reasons (fatigue, lack of initiative, transport, financial means) and provide information about available services within the community. |

Up to 53.8% of people with DM1 report not practicing recreational activities.[223] Another study shows that between 22% and 26% of people with DM1 report that their ability to participate in recreational activities is considerably restricted and more than 24% are unsatisfied with their participation in recreational activities.[15]

**T**he following issues were listed to explain participation difficulties in leisure and sport of DM1 patients: physical limitations (29%), limited financial capacity (28%), fatigue (25%), the distance between their home and recreational centers (18%), activities not adapted to their condition and needs (14%), help is required (14%) and difficulties related to transportation (11%).[104] In another study, the factors that explain the risk of low participation were weakness of lower extremities, fatigue, support from family and friends, access to governmental services and a low level of education.[149]

**NURSING INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guidelines.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Physical condition: Physical activity

| PHYSICAL CONDITION | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| Do you engage in any type of physical activity (for a period of 30 min., three times per week at a moderate intensity)? | Y ☐ N ☐ | If yes, describe the nature, frequency and intensity of the activity:<br>_____<br>_____<br>_____<br>If no, proceed with education about the potential benefits of physical exercise. |

The practice of physical activity is very limited in the DM1 population and about 76% of affected people do not follow recommendations to make 30 minutes of physical activity at least three times per week[104]. The most recent Cochrane review reported that physical activities have not been shown to prevent loss of muscle strength.[228] However, moderately intense strength training in DM1 appeared not to be harmful, but there was insufficient evidence to establish its benefit.[229] As DM1 is a population with high levels of obesity, it is essential to highlight the role of physical activity as part of a healthy lifestyle.

**NURSING INTERVENTIONS**

There is no DM1-specific intervention. Follow national clinical guidelines for physical activity.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Income and tax measures

| INCOME AND TAX MEASURES | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| What is you main source of income?<br>Do you experience financial difficulties?<br><br>Do you receive:<br>• Disability tax credit ? | <br><br>Y ☐ N ☐<br><br>Y ☐ N ☐ | This question is optional as it may be innapropriate in certain countries to inquire about income sources.<br><br>If yes, provide information regarding budgeting ressources and refer patient to appropriate services.<br><br>If no, verify eligibility criteria. |

In 1983, 43.6% of people with DM1 reported reliance on social assistance as compared to 12.2% in the controlled population and two out of five families lived below the poverty line.[230] In 2007, in the same DM1 population, 42.5% still reported reliance on social assistance and 70.5% had an income lower to the median Canadian family income ($48,653).[19]

People with DM1 often have low education, lower cognitive capabilities and are apathetic. The chances of experiencing socioeconomic deprivation are weighted heavily for patients with DM1. Each additional 100 CTG repeats was found to increase the odds of relying on social assistance by about 35% and having low social support by about 22%.[19]

**NURSING INTERVENTIONS**

If a problem is detected, regular support must be offered via budgetary services available in the community.

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Family and social environment

| FAMILY AND SOCIAL ENVIRONMENT | | |
|---|---|---|
| **Evaluation** | | **Reference/Intervention Criteria** |
| • How many members of your family live with you or near your house? | _____ | The questions aim to identify social loneliness.<br>If one of the answers suggests the presence of social isolation, explore the situation with the patient and refer to a social worker. |
| • Do you have friends? | Y ☐ N ☐ | |
| • Are you satisfied with your social life? | Y ☐ N ☐ | |
| • Do you consider that your family or social network offers you adequate support? | Y ☐ N ☐ | |
| • Are you a member of a support group in your community? (Give some examples like Myotonic Dystrophy Foundation, Muscular Dystropy Association ) | Y ☐ N ☐ | If the answer is no, offer information about support groups in the community. |

With regards to geographic location, people with DM1 are more often located in poorer neighborhoods than the reference population (35.4% compared to 24.1% respectively).[16, 19]

The combination of low education, low employment rate, low family income and frequent reliance on social assistance confirms that people with DM1 tend to have a low socioeconomic status and explains the concentration effect in disadvantaged neighborhoods.[19, 227] This residential segregation affects DM1 patients with having the social effects of residing in a neighborhood where the majority of their neighbours are also poor. Such a phenomenon could play a role in the putative perpetuation of poverty in DM1 and contribute to social exclusion and isolation.[14] On the other hand, being an autosomal dominant disease, DM1 can affect several individuals in the same family, which may bring about the phenomenon of a normalization effect. As a result and quite often, DM1 persons, even severely affected, do not perceive a reduction in their social participation because their immediate social environment has a similar level.[231]

**NURSING INTERVENTIONS**

In order to improve their well-being, participation in a support group is suggested for people with a neuromuscular disease.[231, 232]

## SOCIAL PARTICIPATION

### ELEMENT OF SURVEILLANCE
Parental neglect

| **PARENTAL NEGLIGENCE** (observations and clinical judgement) | |
|---|---|
| **Evaluation** | **Reference/Intervention Criteria** |
| During the evaluation process, remain alert to signs that can suggest parental negligence or a difficult parent/child relationship. If in doubt, proceed with a referal to appropriate community services or youth protection services. | _____<br>_____<br>_____<br>_____ |

The prevalence of parental negligence is not well documented although the phenomenon is probably not rare especially when the mother is affected by the childhood or early adult phenotype.

Parents who suffer from the childhood or early adult onset have often difficulty providing emotional support, cognitive stimulation and daily care for their children leading to a situation of negligence. The presence of mental retardation or cognitive disabilities can explain, in part, the situation. Parents who have the adult phenotype often live in a difficult socioeconomic environment that can compromise the development of the child.

**NURSING INTERVENTIONS**

Report about parental negligence should be done according to national laws.

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