Abstract

Oculopharyngeal muscular dystrophy (OPMD) is an autosomal dominant form of late-onset muscular dystrophy. Ptosis (droopy eyelids) and dysphagia (difficulty swallowing) are the most common presenting symptoms. The purpose of this phenomenological study was to describe the experience of living with OPMD. Purposeful sampling was used to recruit individuals with genetically confirmed OPMD who displayed ptosis and dysphagia, were 40 years or older, English speaking, and were willing to consent to the tape-recording of the interviews. An unstructured interview format was used to solicit the participants’ perspectives of living with droopy eyelids, difficulty swallowing, and a genetic disorder. The interviews were audiotaped and transcribed verbatim. Colaizzi’s Method was used to analyze the data, which identified five comprehensive themes. The themes that emerged describing the experience of living with OPMD were “Adjusting to Change”, “Managing Misconceptions”, “Seeking Normality”, “Facing the Future”, and “Informing Children”. The information derived from this study will assist nurses to identify the burdens of living with OPMD and to intervene appropriately early in the course of the disease.

Key words: ptosis, dysphagia, oculopharyngeal muscular dystrophy, genetic, autosomal dominant transmission

Introduction

Oculopharyngeal muscular dystrophy (OPMD) is an autosomal dominant form of adult-onset, progressive muscular dystrophy (Becher Morrison, Davis, Maki, King, Bicknell, et al., 2001; Munitiz, Ortiz, Martinez de Haro, Glover, Ferri, Monotoya, et al., 2003; Urtizberea, 2004). OPMD exists in several clusters across five continents with the largest cluster identified to date in the French-Canadian population of Quebec. The prevalence of OPMD in Quebec has been reported close to 1:1,000 (Urtizberea), which makes the disease as common as Parkinson’s disease and more common than any other types of muscular dystrophy in Quebec (Stimson, 2004). Autosomal dominant genetic transmission is the most common type of OPMD and is exclusive form intrinsic to the French-Canadian population (Brais, 1998). This mode of genetic inheritance dictates that only one faulty gene is needed to cause the disease and that an affected parent has a 50% chance of passing it on to each child. Because disease symptoms are manifested after 40 years of age and usually in the fifth or sixth decade, most individuals are past their child-bearing years when they are diagnosed with OPMD. Thus, there is a 50% chance the disease was unknowingly passed on to their children (Brais).

Individuals with OPMD usually become symptomatic between the ages of 40 and 60 years of age. The symptoms are progressive and, at the present time, the only treatment available is supportive in nature. Ptosis (droopy eyelids) is the most common presenting symptom occurring in 100% of cases with an onset over 60 years of age (Urtizberea, 2004). Ptosis, when severe, becomes problematic because the eyelid may cover the pupil and patients are unable to see without manually lifting the eyelid or tilting their neck backwards causing strain on the extensor muscles of the neck. Most individuals lose part of their visual field and will require surgical ptosis correction. Dysphagia (difficulty swallowing) is usually present and can result in pooling of secretions in the nasopharynx or episodes of aspiration (Stimson, 2004). The most common causes of death in this population are malnutrition and starvation or aspiration and pneumonia (Urtizberea). The purpose of this qualitative study was to describe the experience of living with OPMD.
Review of the literature

Studies in Quebec determined that individuals of French-Canadian descent have the same form of OPMD from the original genetic source. The literature indicates that three Eymard sisters emigrated from France to Quebec in 1648 and introduced OPMD into their lineage for 11 generations that followed (Brais, 1998). In the 1960s, researchers used extensive records of the Roman Catholic Church in Quebec to probe the genealogical origins of this cluster. After evaluating more than 160 individuals with the disease, the common origin of all reported persons of French-Canadian background was established (Brais, 1998).

In 1998, a Canadian team led by Dr. Guy Rouleau, a geneticist at McGill University and Montreal General Hospital, discovered the gene linked with OPMD. The gene encodes a protein called nuclear poly (A)-binding protein 1, (PABPN1), which is located on chromosome 14q. The gene codes for a protein that affects the manner in which cells process RNA, a major step in cells’ manufacturing process. It has been conjectured that the problem in OPMD is not related to a loss of the protein manufacturing process. Brais and Rouleau (2006) have postulated that the protein still carries out the normal function, however, takes on a new, deleterious function in OPMD. PABPN1 is found in almost every cell in the body and is located in the nucleus where DNA is stored. Normally, the PABPN1 gene contains six consecutive repeats of GCG (the three-letter repeated code within the PABPN1 gene sequence of the DNA). However, the PABPN1 gene in individuals with OPMD contains eight to 13 repeats of GCG with GCG-9 (nine GCG repeats) being the most common mutation in the PABPN1 gene. Unlike other triplet-expansion diseases, this mutation is quite stable over generations. In OPMD, extra alanine molecules are incorporated into the PABPN1 protein, which may cause mutated PABPN1 oligomers to accumulate as filament inclusions in the nuclei (Brais, 1998). Repeat expansions of other sizes have been found and may explain why some people have more involved weakness and others only have ptosis and dysphagia. Longer-repeat expansions have been associated with more severe disease progression, as well as weakness in other muscles. While the majority of individuals have one normal and one mutated copy of PABPN1, it is possible to get a “double dose” of defective PABPN1—two mutated copies of the gene (one from each parent). The resulting disease in such cases tends to be more severe and to begin at an earlier age (Brais & Rouleau).

Hermann and Looney (2001) conducted a study that identified the most common symptoms experienced by hospice patients in the last seven days of life, identified interventions used to treat the symptoms, and examined the effectiveness of the interventions. The findings reported dysphagia in 50% of the subjects during the last week of life, which was treated by changing the route of medication administration and education. Missing study documentation, however, limited the ability to evaluate the effectiveness of the interventions. Other studies related to the experience of having dysphagia have been conducted with various other populations including esophageal cancer. A qualitative study conducted by Watt, Centre and Whyte (2003) explored the experience of dysphagia in esophageal cancer and how it impacts quality of life (QOL). The themes that emerged from the data were recognizing dysphagia, the physical experience, the emotions evoked, the impact on social life, and dysphagia and treatment. The authors concluded that dysphagia was a troublesome symptom and affects all aspects of life. Bayer (1984), Wilson (1993), and Liley and Manthrope (2003) examined the experience of receiving long-term enteral feedings. The findings indicated the experience had both a negative and positive impact on aspects of daily life, decision-making, adaptation, loss of control, dependency, body image disturbances, and altered family and social interactions.

Hollman, Ek, Olsson, and Bertero (2004) conducted a study to describe the meaning of QOL in patients with a genetic disease, familial hypercholesterolemia. Findings of this qualitative study noted that for this population, QOL was related to maintaining or regaining harmony in life.

In summary, a review of the literature revealed a dearth of research studies conducted on ptosis, dysphagia, and the OPMD population. Major gaps in describing the experience of people with ptosis, dysphagia, and genetic disorders are apparent. The literature review indicated that relatively little is known about the personal experience of having droopy eyelids, dysphagia, and a genetic disorder. This phenomenological study was aimed at building the knowledge base related to living with OPMD.

Purpose

The purpose of this research study was to describe the experience of living with OPMD from the participants’ perspective. The following research questions were explored:
1. What is the experience of having droopy eyelids?
2. What is the experience of having difficulty swallowing?
3. What is the experience of having a genetic disease?

Table 1. Participant demographics

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**Method**

**Design**
Phenomenology, a qualitative research tradition, was used to describe the experience of living with OPMD. Phenomenology arises from a naturalistic paradigm that indicates many realities may exist regarding an experience. This study was designed to uncover the meaning of the human experience of having droopy eyelids, difficulty swallowing, and a genetic disorder from the participants’ perspective (Fain, 2004).

**Participants**
Purposeful sampling was used to recruit 10 individuals (five males and five females) ranging in age from 49 to 73 years (Table 1).

Inclusion criteria were individuals diagnosed with OPMD who displayed ptosis and dysphagia, were 40 years or older, English-speaking, and willing to consent to tape-recording of the interview. If the person did not have genetically confirmed OPMD, he/she was excluded from the study because they may have had a neuromuscular disease other than OPMD.

**Data collection**
Institutional Review Board (IRB) approval was received from York College of Pennsylvania and the University of Western Ontario, London, ON. Participants were solicited from a neuromuscular clinic in central Canada that serves a population of approximately 40 individuals diagnosed with OPMD. The individuals were contacted by phone, asked to participate and, if they agreed, were scheduled for an interview. Written information about the date, time, and place where the interview was scheduled was mailed to individuals who agreed to participate in the study. Participation was voluntary and informed consent was obtained from participants. Any questions or concerns voiced by a participant were addressed and answered satisfactorily before the interview began. The researcher reinforced that participants could stop the interview at any time. The participants’ identities were protected by using a coding number on the audiotapes and transcribed records that were known only to the researchers. Coding numbers ensured privacy, confidentiality, and anonymity for all participants. All audiotapes and transcribed records were stored in a locked cabinet and were destroyed at the completion of data collection and transcription. Pseudonyms were used in all transcripts and documents.

The interviews were conducted either in the individual’s home or in a small, private conference room at the clinic. An unstructured interview format was used to provide greater latitude in answering the questions. The interviews were conducted by one or both of the investigators who have previous experience conducting qualitative research studies. Each interview lasted between 30 and 60 minutes.

**Data analysis**
Bracketing of prior knowledge and the independent analysis of the interviews by both investigators helped to ensure pure description of the data. The transcripts were read and reread to allow the themes of the participants’ experiences of living with OPMD to emerge. Data collection was completed when saturation occurred and no new themes evolved.

Data analysis was conducted based on Colaizzi’s Method for phenomenological research (Fain, 2004; Speziale & Carpenter, 2003). This method of analysis includes the following steps:
1. Describe the lived experience under investigation.
2. Collect the participant’s description of the lived experience.
3. Read all the participants’ descriptions of the lived experience.
4. Extract significant statements and articulate the meaning of each.
5. Organize the statements and meanings into themes.
6. Write a narrative description.
7. Return to the participants for validation of the description (member check).
8. If new data emerge, incorporate them into the final description.

**Trustworthiness of the data**
Several procedures are used in phenomenology to ensure the themes that emerge are accurate and consistent representations of the experience as described by the participants. In this study, both investigators read the transcripts and agreed on the themes to ensure credibility. Further validation of the themes was achieved by member checks. The findings were shared with the participants who validated the themes.

**Findings**
The results indicated that all participants suffered both physiological and psychosocial burdens related to living with OPMD. Five comprehensive themes emerged from the data that described the essence of this experience:

a) Adjusting to change
This theme expresses the physiological toll that droopy eyelids had on the participants. As the eyelids began to droop and affected their eyesight, participants responded by putting their chin in the air and arching their backs in order to see. Several participants complained of neck and shoulder pain due to pressure placed on the cervical musculature resulting from keeping their chins in the air and arching their necks. In a few cases, the participants noted their walking became unsteady and often would need to arch their lower back in an attempt to improve their eyesight. The following examples taken from the interviews represent this theme:

Nancy: Because you are looking out from underneath your eyelids, you tend to raise your chin.

John: I would actually manually prop my eye open with my fingers.

Mary: I developed pain in my neck and arm from arching my neck. That was when I knew that I needed to see a doctor.

Anita: When my eyes started to droop and I was wearing my glasses, all I could see was my eyelashes, which were magnified because of the bifocals. I used to read a book every two days, but now I read nothing but the newspaper occasionally.

All of the participants had frontalis sling surgery to correct the ptosis, which was perceived as a positive experience that improved eyesight and eliminated the neck and shoulder pain in all cases.
b) Managing misconceptions
This theme illustrates one of the psychological issues encountered. Participants frequently experienced unkind comments from people they didn’t know with regard to the ptosis. Although the remarks were not meant to be offensive, they did result in sorrow and depression in several cases. Examples of quotations that represent this theme included:

Linda: People would ask if I was tired or ever once when I stopped to cross the border in the United States, I was evaluated to see if I was intoxicated and driving under the influence of alcohol.

Mary: I am a nurse and was at a lecture one day. The woman I was with accused me of sleeping during the program.

Bob: I always liked to play golf. When my eyes started drooping, the other guys would give me a hard time about falling asleep in the golf cart. I finally had to stop playing because my eyesight was affected and I couldn’t open my eyes enough to see where I was hitting the ball.

All the participants described incidents of being embarrassed because of their droopy eyelids. Such feelings resulted in several individuals becoming reclusive until they had corrective surgery. Following the surgery, they were more comfortable and returned to participating in social activities they had avoided because of unkind comments.

c) Seeking normality
This theme indicates the profound impact that dysphagia had on the participants’ health, nutrition, and socialization related to the normal process of eating. Although friends and family were supportive of their difficulty in swallowing, the following examples demonstrate the challenges that participants experienced:

Susan: We eat very slowly. It’s kind of a standing joke. There are four of us girls with the disease and if we go out for lunch, count on an afternoon because we don’t talk and eat at the same time or we choke.

Linda: I eat before I go out with my friends. Then I have tea or coffee while they are eating. That way I can still visit with my friends, but I can be comfortable and not worry about choking.

Harry: The people I associate with, they know that I take forever to eat. So, I mean, that’s an accepted fact. We know what I can eat and what I don’t eat. People have to wait to clear off the table because I’m going to be the last one there.

The dysphagia had a significant impact on the social process of eating and enjoying a meal with friends and family. Each person described in detail what foods they avoided and which foods they would eat when they were in a social setting. All of the participants had learned adaptive behaviours to cope with eating both at home and in restaurants. Several individuals had surgery, cricopharyngeal myotomy, which improved their swallowing for limited periods of time. Two individuals had aspirated several times and lost significant amounts of weight. This resulted in the individuals having feeding tubes inserted for nutritional support and the prevention of aspiration pneumonia. Although the feeding tubes had an impact on their nutritional status, they continued to experience psychological stress with respect to the inability to eat normally.

d) Facing the future
This theme represents the pain of not knowing what the future holds related to progressive difficulty swallowing and the ongoing fear of choking that resulted in both physiological and psychological stressors. The individuals described fears related to aspiration, malnutrition, and dying from choking. Here are examples of this theme:

Nancy: One of my other fears is who will help me in the future—puree the food or assist with the feeding tube? My husband’s health is not that great right now. So, I am looking at the possibility of being alone.

Susan: I have had two episodes in the last six months where my husband actually had to do the Heimlich manoeuvre. That was really scary.

Betty: I have two uncles that got feeding tubes. One of them didn’t want to use the tube, so he uses it for breakfast. But then he eats lunch and dinner after that. I hope I don’t have to make the decision between my quality of life, not being able to eat, or having to depend on a tube.

Surgical intervention does not correct the dysphasia, but can improve the underlying problem for a limited time. Participants were well-aware of the high risk for complications related to the difficulty swallowing. They expressed their concerns about the future and its consequences. However, most had not made any decisions about what treatment they would accept when the choking became life threatening.

e) Informing children
This theme broadens the knowledge we have regarding the very different reactions individuals had when they learned they had a genetic disease. All of the participants struggled with the psychological impact they experienced when divulging the diagnosis to their children. The following examples demonstrate the various reactions that were verbalized in the interviews:

Doris: It was hard telling my four children. They all want to be tested. My one daughter was thinking of having another baby, but now she doesn’t want to get pregnant until she gets tested and makes sure she doesn’t have the gene.

Bob: My children were informed, but they chose not to be tested. It is not something they want to deal with right now.

Susan: I don’t feel guilty and I don’t feel angry. I feel, I feel bad. Because, well, I didn’t know I had this disease and it’s too late. I have two teenage daughters and neither of them wants to be tested until they have symptoms.

Betty: I wouldn’t wish it on my kids for all the tea in China. I hope that they will avoid it, but I don’t know.

This theme describes the intense psychological struggle the participants experienced when they discussed their illness and its implications with children. For all of the participants, this experience was harder than the news of their own diagnosis.
Discussion

People who are living with OPMD felt there were negative aspects to their lives related to having this disease, which included both physiological and psychosocial stressors that were common in their daily lives. Most people had learned adaptive behaviours and sought medical intervention to address the droopy eyelids and the difficulty swallowing. Their hopes focused on maintaining their health and well-being as long as possible. Many expressed thoughts of “living for today”, “making the most of their situation”, and “there are people who are worse off than I am.” There were mixed emotions and feelings regarding knowing they had a genetic disease that has been, or could be transmitted to their children. For those individuals whose children had been tested and were positive for the gene, it was “more hurtful having the disease myself.” The underlying supposition was that they had to “be strong for their family.”

Limitations of the study included the presumption that only French-Canadians with OPMD would be included in the study. Participants of other ethnic backgrounds, however, were interviewed and results suggest the experience of living with OPMD is not affected by ethnicity. While purposeful sampling results in a selection bias, this approach was necessary to ensure participants had first-hand knowledge of this experience. The small sample size precludes generalization of the findings to other populations and geographical settings.

Future research of interest could include studying cultural differences of this experience by replicating the study with other ethnic groups such as the Hispanic group in New Mexico, Bukhara Jews living in Israel, and persons of Asian descent. Exploring QOL issues with various ethnic groups would provide additional data related to the experience of living with OPMD.

Summary

The findings of this study indicate that health care workers need to understand what it means to live with OPMD and how it affects the person’s life. They need to offer support on how to manage both the physiological and the psychosocial burdens of this experience. The knowledge related to the experience of living with OPMD will assist health care workers to identify these burdens early in the course of the disease and offer appropriate interventions to the individuals.

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