Psychosocial Issues That Face Patients With Charcot-Marie-Tooth Disease: The Role of Genetic Counseling

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Charcot-Marie-Tooth (CMT) disease is a hereditary debilitating progressive muscular atrophy and sensory neuropathy of the distal extremities. CMT is usually nonlife threatening. Signs of the disease usually present in childhood or in young adulthood and the level of disability can be variable within and between families. Research addressing specific psychosocial and emotional issues faced by individuals with CMT is limited. Fourteen adults with a clinical and/or molecular diagnosis of CMT (ages 32–74 years) consented to an audio taped interview. The format of the interview was based around an informal questionnaire to prompt and guide the interviewee to describe their experiences of living with a disabling genetic disorder. The interviews focused on their experiences of first symptoms and diagnosis, their life experience with CMT, their limitations due to disability and the role of genetic counseling. This study identifies and explores life issues that individuals with CMT may face, specifically grief over the loss of independence, emotional pain and stress such as embarrassment and guilt of passing on a gene mutation, impact on quality of life, the impact of wearing orthopedic devices, and fear of progressive disability. Our findings suggest that there are emotional and psychosocial issues specific to affected individuals at different life stages and genetic counselors need to be aware of these issues in order to provide age appropriate support and advice to individuals affected by CMT.

KEY WORDS: Charcot-Marie-Tooth disease; genetic counseling; psychosocial issues; physical disability; quality of life; progressive disorder.

INTRODUCTION

Genetics of Charcot-Marie-Tooth Disease

Hereditary motor-sensory neuropathies (HMSNs) are a group of slowly progressive neurological disorders affecting the motor and sensory components of the peripheral nervous system. Charcot-Marie-Tooth (CMT) disease is an HMSN where the nerve dysfunction caused by the disease results in sensory abnormalities and wasting and weakness of distal muscles, particularly of the lower limbs. CMT is the most common inherited peripheral neuropathy, affecting approximately 150,000 Americans. There is a wide range of age of onset, severity and rate of disease progression even within a given family. People with CMT may present with features which may not be obvious to other individuals (e.g., they may develop loss of sensation on the soles of their feet or of vibratory sensation). The purpose of genetic counseling is to provide essential information to the patient and family about the genetic nature of the disease, inheritance patterns, and the potential risk to future generations. Genetic counseling is a specially trained health care professional who uses communication and decision-making skills to help individuals and families understand and adapt to genetic conditions.
in hands and feet, muscle cramps and muscle pain) and they can also develop gait problems, foot deformities (high-arched feet and flat feet), claw hand deformity, scoliosis or kyphosis, diaphragm and/or vocal cord paralysis, loss of balance and manual dexterity, leading to progressive loss of mobility, and independence. (Parry, 1995; Garcia, 1999; Lovelace, 1999; Thomas, 1999; Scriver et al., 2001).

CMT is genetically heterogeneous, and can be inherited in an autosomal dominant, autosomal recessive, or X-linked dominant fashion. Autosomal dominant inheritance is the most commonly observed pattern of inheritance accounting for 80–90% of CMT (Bird, 2004). CMT is divided into two subtypes based on pathological and electrophysical findings, CMT Type 1 and CMT Type 2 (Dyck and Lambert, 1968). CMT1 (also known as HMSN I) is the demyelinating form and is characterized by median motor nerve conduction velocities (NCVs) that are reduced to <38 ms⁻¹ (normal >40–45 ms⁻¹). An “onion bulb” formation is often seen on nerve biopsy. CMT2 (also known as HMSN II) occurs when there is axonal degeneration and is characterized by preserved or slightly reduced NCVs and the absence of “onion bulb” formation (Hanemann and Muller, 1998; Lovelace, 1999; Scriver et al., 2001; Bird, 2004). The majority of HMSN I cases (70–80%) are designated CMT1A and have a dominantly inherited 1.5 Mb duplication on chromosome 17p11.2, which contains the peripheral myelin protein gene (PMP22) (Bird, 2004). A review of all genes identified in CMT is beyond the scope of this paper. Readers are referred to www.geneclinics.org for a full description of all identified CMT genes (Bird, 2004).

As molecular genetic testing in the clinical setting is not available for all the currently known genes that underlie CMT, confirmation of the diagnosis of CMT is often based on a combination of family and medical history, physical examination, electrophysiology (EMG) and NCV testing, and occasionally nerve biopsy (De Jonghe et al., 1999; Athena Diagnostics, 2002; Bird, 2004). It is important to distinguish CMT from other genetic neuropathies (e.g., amyloid neuropathy) or acquired causes of neuropathy that may be treatable (e.g., alcoholism and vitamin B12 deficiency), and neuropathies associated with chronic disease (e.g., diabetes mellitus). Although differentiating between CMT subtypes currently has no therapeutic consequences, identification of a causative mutation in an affected individual gives the individual or couple the option of prenatal diagnosis and/or preimplantation genetic diagnosis (PGD).

Disability and the Impact of CMT on Day-to-Day Living

There is variation in age of onset, severity and rate of disease progression in different CMT subtypes, which makes accurate prognostication challenging. The medical view of the relative severity of CMT varies. For example, in one paper, 8 out of 23 (35%) clinicians interviewed said CMT is “serious but not lethal,” and 15 out of 23 (65%) claimed CMT is “not serious” (Wertz and Knoppers, 2002, p. 32). Other studies have categorized CMT as a “relatively mild” disease; while one study states 20% of patients they observed were “severely disabled” (De Jonghe et al., 1997; Reilly, 1998). It is clear that there is variability in severity within and between families and thus it is not appropriate to generalize as to whether this disorder is “mild” or “severe.” This is highlighted by Wertz and Knoppers (2002) who found that there is “not sufficient consensus amongst experienced genetics professionals to define serious genetic conditions.”

Miller et al. state, “We believe that bodies define us, they are who we are. How good we look and how well we move are foremost in our evaluations of ourselves and others” (p. 123). A disability, whether mild or severe, can significantly impact a person’s day-to-day living. Most individuals need to adapt alternate ways to function in everyday life, for example employing the use of a wheelchair, employing a home carer and/or making home adaptations (remodeling the home to include a ramp) (Miller & Simmons, 1999). Richards (1994) found that individual perceptions of disability and diagnosis depended on the individuals’ personal experiences of the disease, personality, family and social support networks, and also on the previous generations’ experience of the disease. Marshak et al.’s (1999) investigation of issues affecting families coping with disability in a general sense identified that adjustment to the disability is influenced by family structure, accessibility of resources, cultural beliefs, personal beliefs and social support.

These findings imply that for individuals affected by CMT, it is important to explore all these issues and identify specific individual issues so that genetic counselors may assist their clients with developing their coping strategies.

Psychosocial Issues for CMT Patients

The emphasis of the impact of CMT on affected individuals differs between the medical and support group literature. The focus in the medical literature