Hereditary Motor Sensory Neuropathy: Understanding Function Using Motion Analysis

Sylvia Õunpuu and Kristan Pierz

Abstract

Hereditary motor sensory neuropathies, or Charcot-Marie-Tooth disease, represent a heterogeneous group of inherited neuropathies that are characterized by progressive wasting and resulting weakness of the distal muscles in the legs and arms. Lower extremities are typically initially effected and, as result, impact ambulation. At present there is no curative treatment available; therefore, treatment of gait issues is often sought to help with ambulation and activities of daily living. Computerized motion analysis techniques have improved our understanding of the various presentations of hereditary neuropathies and can assist in making optimal treatment decisions to improve gait. These presentations include three distinct ankle variations: excessive equinus (toe walking), cavo-varus (lateral border weight bearing), and flail foot (heel weight bearing) patterns. As each patient presents differently in terms of deformity specifics and severity, a detailed analysis that describes ankle/foot function during gait in terms of foot pressures, muscle activity, kinematics, and kinetics along with clinical examination information such as muscle strength and passive range of motion is very beneficial. Assessment of treatment outcomes from bracing to orthopedic surgery as well as disease progression which also varies person to person is necessary to develop evidence-based treatment indications and goals. Motion analysis can play a very important role in the assessment of inherited neuropathies on both an individual patient basis and in research with the ultimate goal of improving treatment outcomes.

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Keywords

Hereditary neuropathies • Charcot-Marie-Tooth • Clinical gait analysis • Peak dorsiflexion • Joint kinematics • Joint kinetics • Ankle

Abbreviations

AFO	Ankle foot orthosis
CMT	Charcot-Marie-Tooth
EMG	Electromyography
PLS	Posterior leaf spring orthosis

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Introduction

Hereditary motor sensory neuropathies, or Charcot-Marie-Tooth disease (CMT), represent a heterogeneous group of inherited neuropathies that are characterized by progressive weakness and resulting wasting of the distal muscles in the extremities (legs and arms). This group of neuropathies was described in detail with the tools available at the time in 1886 by neurologists Charcot, Marie, and Tooth who defined them as peroneal muscular atrophies of familial origin (Charcot 1886; Tooth 1886). These neuropathies are length dependent, meaning the longest nerves in the body are affected, and therefore, the lower extremities are typically initially affected in the course of disease progression. In terms of function, the resulting muscle weakness results in foot deformity such as flatfoot or high arch and gait issues such as ankle instability, clumsiness, and slow running. During standing and walking, ankle support is often needed and provided by orthoses, and in some cases walking becomes so difficult over time that wheelchair mobility is required. At present there is no curative treatment available, so treatment of gait issues can help provide more functional ambulation and improve/facilitate participation and activities of daily living. As with any complex movement disorder, CMT presents differently in each patient in terms of deformity, severity, and rate of disease progression. Comprehensive motion analysis techniques can provide objective documentation of (a) the pathomechanics, (b) disease progression, and (c) treatment outcomes. The goal of this chapter is to highlight what we have learned about the pathomechanics of gait in persons with CMT using comprehensive motion analysis techniques. The application of motion analysis is in its infancy for this gait pathology. The ultimate

goal is to improve treatment outcomes and to understand prognosis for future function near the time of diagnosis through a better understanding of gait pathomechanics. Although progress has been made, there is much more to be done.

State of the Art

The application of motion analysis in this clinical setting is relatively new in comparison to other gait disorders such as cerebral palsy and spina bifida, so our understanding of gait function in persons with CMT limited. Because CMT is a rare disease, the clinical application of motion analysis in this patient population is not as common, and research applications for the most part have focused on small diverse groups of patients that span a wide age range. However, over time this body of literature has grown, and clinicians have gained more understanding of the pathomechanics of gait. The presentation of CMT in classical textbooks generally describes a common set of impairments (weakness and contracture) that result in a specific gait pattern including excessive equinus in swing and increased hip flexion, pelvic hiking, and circumduction to clear the foot (Holmes and Hansen 1993). Clinical experience shows that many patients with CMT do not exhibit excessive equinus in swing and the associated proximal compensations. Also cavo-varus foot deformity is noted as common (Smith 2002); however, many patients with CMT have flail feet with pes valgus. It is clear from previous research that incorporates comprehensive motion analysis assessments that there are substantial variations in presentation and severity of impairments (Burns et al. 2005, 2006; Garcia et al. 1998; Vinci and Perelli 2002; Vinci et al. 2006) that result in a variety of gait pathologies (Don et al. 2007; Kuruvilla et al. 2000; Newman et al. 2007; Ramdharry et al. 2009). As a result, this latter group of authors divided CMT patients into groups based upon gait pathology. In many cases gait features often overlapped, that is, multiple features were present in a single patient. These studies mostly focused on adults with CMT, and as a result there is very little understanding of CMT gait pathology evolution during childhood when the impairments typically present. However, there are a few exceptions. The first signs of CMT were evaluated by Burns et al. 2009 and include issues with ability to toe walk and run and clumsy gait (Burns et al. 2009). Ferrarin (Ferrarin et al. 2012) divided gait pathology into three categories in a group of children and adolescents using motion analysis outcomes: (a) pseudo normal, (b) drop foot only, and (c) foot drop and push-off deficiency. In a group of similarly aged patients, Ounpuu (Ounpuu et al. 2013)focused on peak dorsiflexion in terminal stance as a way to differentiate gait in persons with CMT as this particular gait parameter is relevant to treatment strategy. In this study, patients were placed into three groups: (a) less than typical peak dorsiflexion, (b) typical peak dorsiflexion, and (c) increased peak dorsiflexion. Patients with CMT, however, rarely present with one issue at the ankle, so in groups (a) and (c), there was also simultaneous increased equinus, and in groups (b) and (c), there was delayed peak dorsiflexion in stance. There is, however, still work needed to further refine gait classifications in these patients, understand disease progression and implications of asymmetry and

ultimately link gait issues to phenotype. Initial reports on CMT often depicted large asymmetries, however, it has been recently reported that asymmetry is not that common in CMT in terms of a wide variety of outcome measures including gait parameters (Burns et al. 2012).

In that there is such variation in presentation, treatments need to be patient specific and based on a comprehensive patient examination including motion analysis especially at the level of the ankle and foot (Jani-Acsadi et al. 2015). There is, however, very little objective documentation of treatment outcomes in terms of gait function using comprehensive motion analysis techniques. As more centers incorporate comprehensive motion measurement techniques as part of the standard of care, this will become possible.

Gait Function and Treatment Options

This section will include a description of the following for persons with CMT: (a) the current understanding of gait pathology in terms of gait analysis parameters and associated clinical examination findings and (b) the current standard of treatment for gait-related issues including therapies, orthoses, and orthopedic surgery. Example patient data will be incorporated to illustrate common findings.

Clinical Examination Findings in CMT

Clinical examination findings which are collected using standard protocols for the assessment of strength, passive range of motion, and bony deformity are integral to the interpretation of motion analysis data so need to be a part of a full assessment of gait pathology. Clinical findings help to determine the primary impairments that lead to gait pathology. Persons with CMT typically have impairments that include weakness, contracture, and bony deformity which are further complicated by the disease progression (increased problems over time) as well as issues with reduced sensation and reflex response which also need to be assessed. For the context of this chapter with the focus on gait analysis, we will briefly discuss the primary impairments that impact gait in persons with CMT.

The first manifestations of disease for most persons with CMT are distal limb weakness and muscle atrophy. Sensory loss and absent reflexes are also present (Thomas 1999). Comprehensive research on strength testing methods and comparison between controls and persons with CMT1A in terms of strength and passive range of motion findings have been completed by Burns and colleagues (Burns et al. 2005, 2009; Rose et al. 2010). Their detailed studies have led to some primary findings in children with CMT and cavus foot deformity which include reduced strength in all of the muscles of the feet. Initial deficits in young children in comparison with their age-matched peers start with weakness of the plantar flexors, dorsiflexors, and evertors of the ankle (Rose et al. 2010). Young children with CMT also have greater inversion-to-eversion and plantar flexion-to-dorsiflexion strength

ratios and a high correlate between dorsiflexion range of motion and foot and ankle strength. Those with CMT also have significantly less passive ankle dorsiflexion range of motion. Strength assessments in adult persons with CMT have also documented weakness in the ankle dorsiflexors, plantar flexors, and evertors (Don et al. 2007; Newman et al. 2007). Strength and range of motion deficits result in gait pathology and should be linked to the interpretation of gait data to determine causes of gait pathology. This has been done in the majority of research involving motion analysis and CMT. Longitudinal data that would help to explain the pathogenesis and progression of this disease in terms of gait decline over time would be extremely helpful and is not yet available.

Gait Findings: As Defined by Motion Analysis Parameters

Motion analysis has led to a more comprehensive understanding of the pathomechanics of gait in persons with CMT. The following section will describe CMT in the context of the following components of a comprehensive motion analysis: gait kinematics and kinetics, electromyography, and pedobarography. Each component provides a unique source of information not available using standard assessments and when combined provide a comprehensive view of the pathomechanics of gait in this population. Motion analysis also provides a unique opportunity to assess gait function objectively over time and to evaluate treatment outcomes such as orthoses and orthopedic surgery.

Kinematic and Kinetic Findings in CMT

As mentioned above, the majority of the previous research focused on CMT and gait includes joint and segment kinematic and joint kinetic outcomes. These data provide an opportunity to better understand the pathomechanics of gait in CMT and help to make links between the primary pathology and compensations noted at proximal joints. It is clear from these works that CMT with its heterogeneous genetic makeup also has a heterogeneous impairment and associated functional presentation. Ankle function during gait in CMT is impacted depending on the impairments of weakness and contracture in the associated musculature. Therefore, presentation can be differentiated by the degree of peak dorsiflexion in terminal stance (which is impacted by both ankle plantar flexor range of motion and strength) and divided into three groups: (a) increased and delayed peak dorsiflexion, (b) delayed peak dorsiflexion, and (c) increased plantar flexion (Ounpuu et al. 2013). Similarly, ankle dorsiflexion in mid-swing and at initial contact is often impacted by ankle dorsiflexor strength but is also impacted by plantar flexor tightness that does not "allow" the anterior tibialis to effectively dorsiflex the ankle in a non-weight-bearing position (swing phase). So CMT commonly presents with excessive equinus in mid-swing and initial contact. Cavus foot deformity is also common but not well documented using current motion analysis methods. As it impacts the available length of the ankle plantar flexors, it is an important consideration for the overall function of the foot and ankle.

Generally, the foot and ankle in CMT fall into three groups of impairments.

- The flail foot with significant weakness in all ankle muscles that leads to increased and delayed peak dorsiflexion in stance and equinus in swing with medial and lateral instability resulting in pes planovalgus deformity over time (Fig. 1a–c). Although claw toes are common in earlier phases of the disease, in this group, the toe flexors are typically weak with no toe contact during stance.
- The cavus foot deformity with lateral weight bearing during stance and delayed peak dorsiflexion in terminal stance consistent with ankle plantar flexor weakness and typically normal or only minimal increased equinus in swing (Fig. 2a–c). The cavus deformity limits available plantar flexor length which when at its end range may mask ankle plantar flexor weakness.
- Equinus ankle deformity with increased equinus in stance and swing due to lack of plantar flexor range of motion (assessment of shank vs. plantar aspect of the foot) (Fig. 3a–c). This lack of passive range of motion may be a result of a

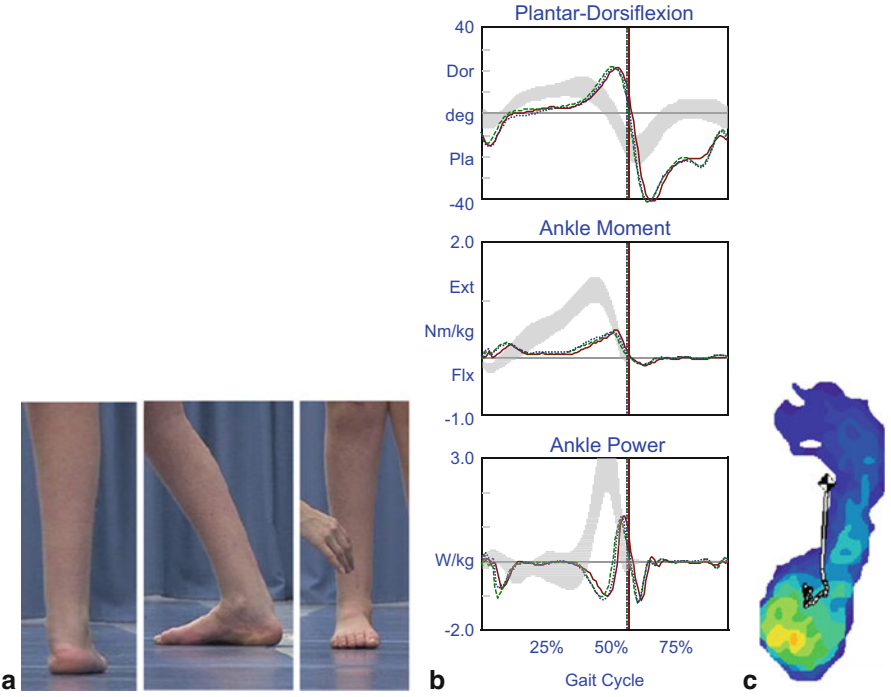

Fig. 1 Example of the flail foot during (**a**) relaxed standing with significant weakness of the ankle musculature as evident in the (**b**) ankle sagittal plane kinematic, moment and power (three gait cycles) during stance and swing phases and in (**c**) foot pressures for the right foot. Increased and delayed peak dorsiflexion, reduced peak ankle plantar flexor moment and power generation in terminal stance and reduced pressures under the distal foot with a limited length of center of pressure path are evidence of ankle plantar flexor weakness. Increased plantar flexion in swing is evidence of ankle dorsiflexor weakness

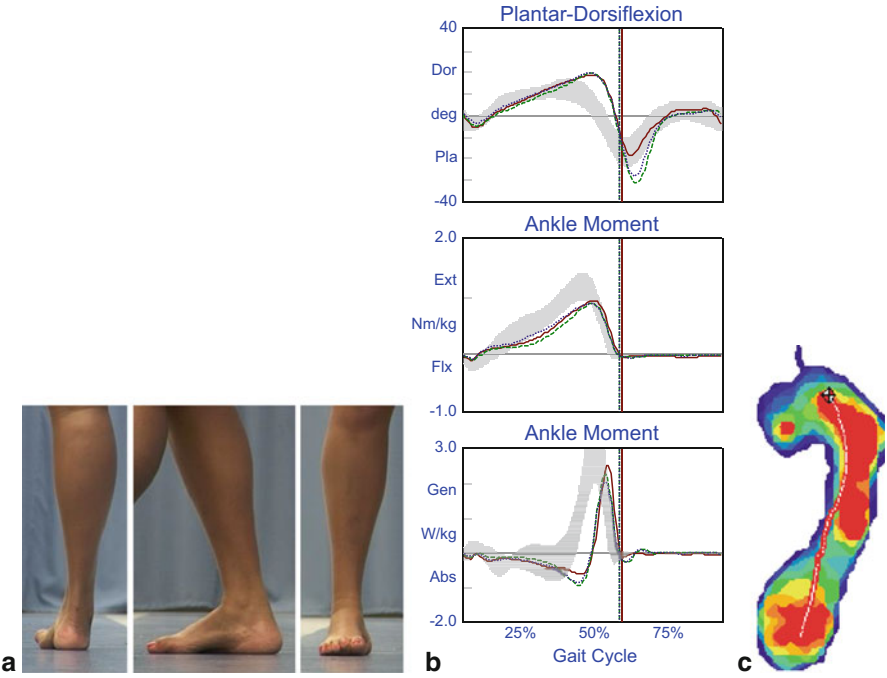

Fig. 2 Example of the cavo-varus foot during (**a**) relaxed standing with some evidence of plantar flexor weakness in the (**b**) ankle sagittal plane kinematic, moment and power (three gait cycles) during stance phase and evidence of lateral weight bearing in (**c**) foot pressures for the right foot. Weakness of the ankle plantar flexors is most evident in the delayed and increased peak ankle dorsiflexion in stance, however, this is not yet manifested at the joint kinetic level

combination of cavus and/or limited plantar flexor range of motion. The cavus deformity limits available plantar flexor length and may mask the presence of ankle plantar flexor weakness.

The compensatory proximal gait findings depend on the ankle impairment. For those patients with increased equinus in swing and dorsiflexion in stance, compensatory steppage gait is common with increased hip flexion and in some cases circumduction to aid in clearance (Fig. 4). For those with increased ankle dorsiflexion in stance, increased knee and in some cases hip flexion occur. Because the kinematic and kinetic presentations vary from patient to patient, the treatment options need to be specific to the patient and are discussed in the Management section below.

Electromyographic Findings in CMT

Muscle activity for those muscles that are used to support the ankle during gait in persons with CMT is often abnormal and is manifested in multiple ways including: (a) reduced activity, (b) no activity, and (c) atypical recruitment patterns. Lack of muscle activity during the appropriate phases of the gait cycle can result in ankle

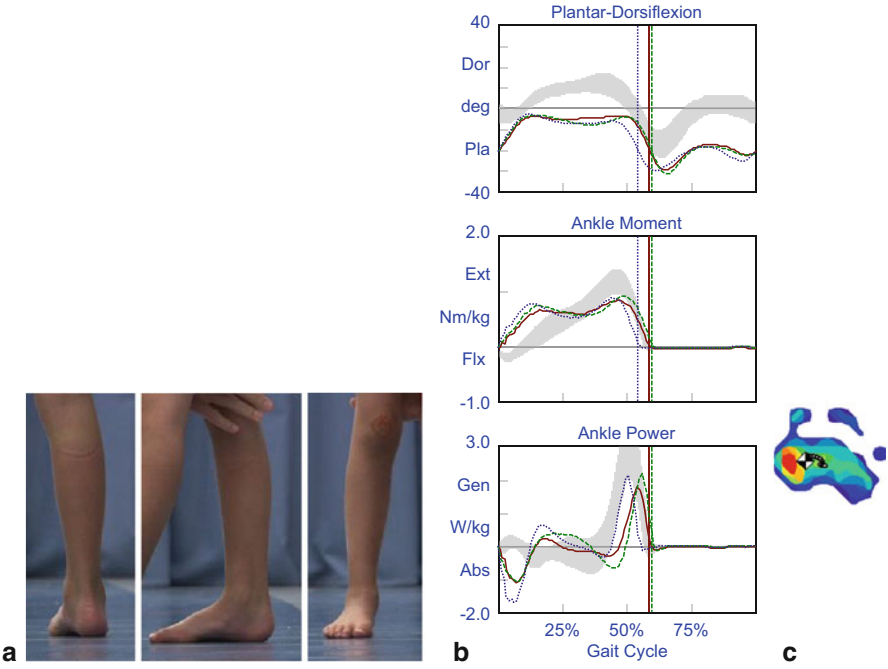

Fig. 3 Example of the equinus ankle during (**a**) relaxed standing with inability to dorsiflex above neutral and increased equinus documented in the (**b**) ankle sagittal plane kinematic, moment and power (three gait cycles) during stance and swing phases and in (**c**) foot pressures with the right forefoot and toes only in contact with the ground instance. Increased plantar flexion in stance and swing is evidence of plantar flexor tightness

lateral instability if the peroneals are involved, increased equinus in swing if the anterior tibial group is involved, and increased peak dorsiflexion in stance if the plantar flexors are involved. Abnormal muscle contractile patterns such as single motor unit recruitment and fasciculations (repetitive single motor unit contractions at rest) are also characteristic of demyelinated muscle that can be noted on the surface EMG signal. Dynamic electromyography (EMG) techniques can provide insight into which muscles are active during which phases of gait and assist in understanding potential for muscles to provide dynamic support when transferred. As in the varied kinematic presentation, one would expect that the muscle function for this patient group is also different from person to person as muscle activity is the driver of motion and stability. So EMG patterns for key ankle muscle groups can show minimal issues or provide evidence to explain why the ankle joint is unstable (Fig. 5a–c).

Pedobarography (Foot Pressures) Findings in CMT

The analysis of foot pressures during gait provides an opportunity to understand where the peak pressures are under the foot and the path of the center of pressure during the stance phase of gait. High mean and peak pressures and pressure-time

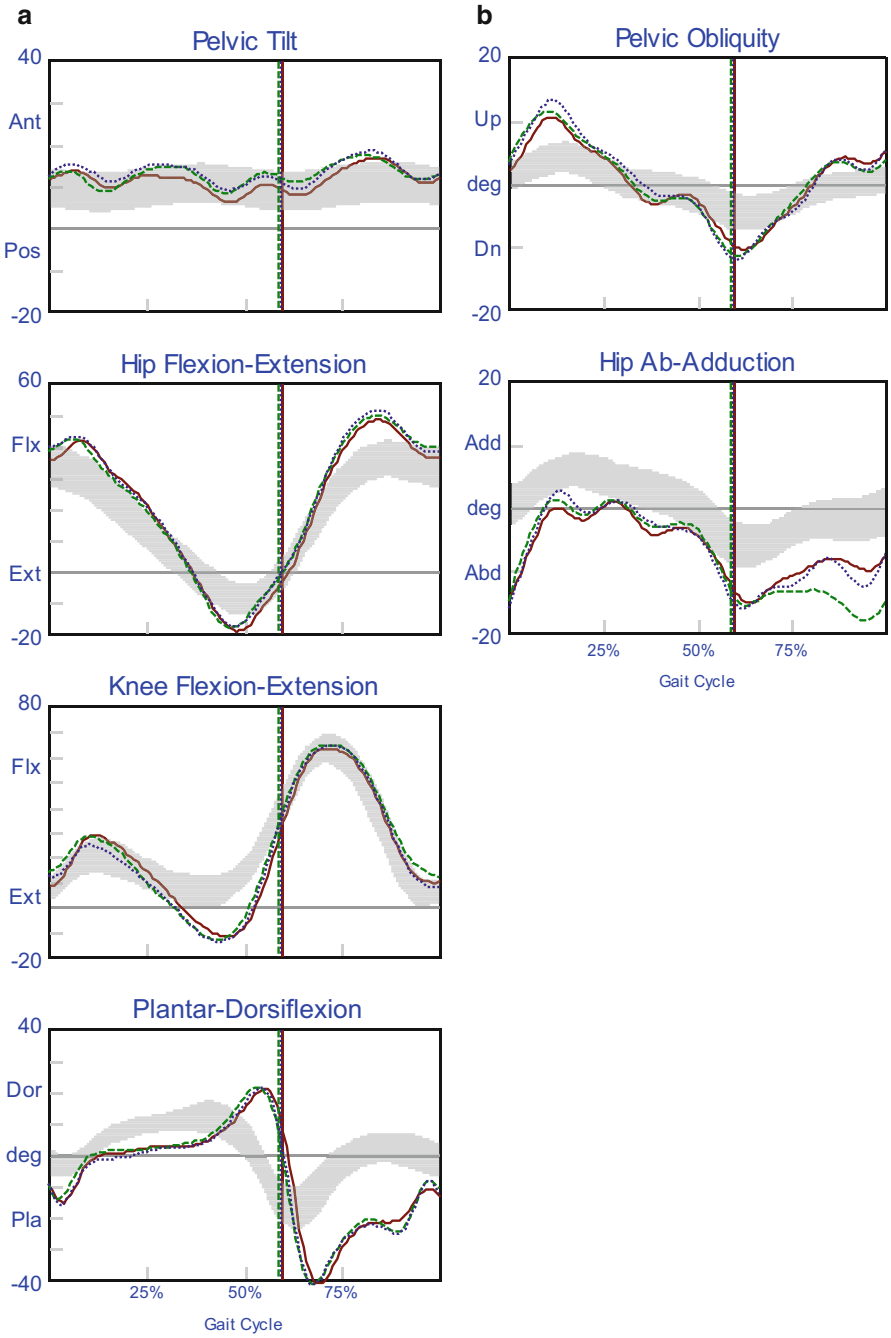

Fig. 4 Compensations for excessive equinus in swing typically include (**a**) increased hip flexion in the sagittal plane (left column second plot) and (**b**) circumduction and pelvic hiking in the coronal plane (right column)

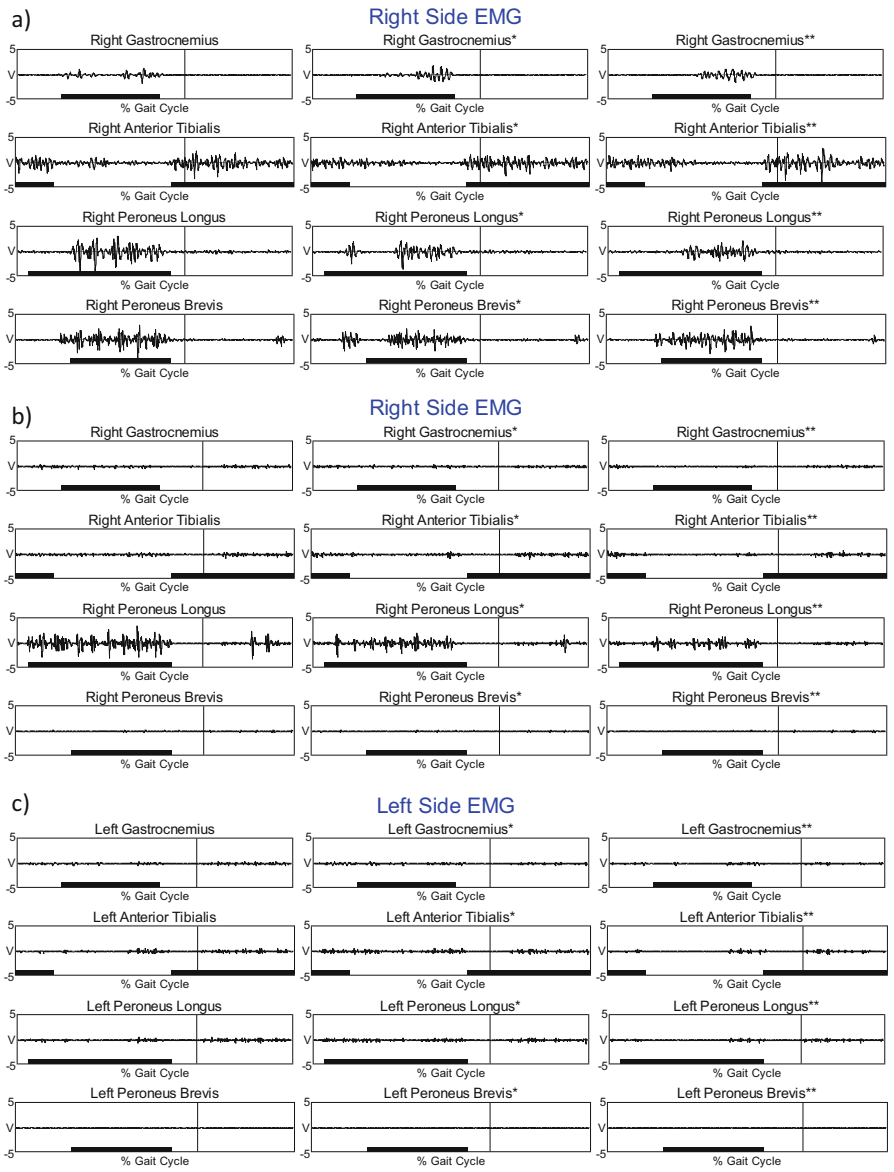

Fig. 5 Example EMG data for three different patients with CMT show heterogeneity of this disease in terms of the primary muscles at the ankle with (**a**) minimal atypical findings except for the gastrocnemius which shows delayed onset and minimal activity consistent with the common finding of plantar flexor weakness and increased dorsiflexion in terminal stance, (**b**) minimal activity other than fasciculations of peroneus longus and (**c**) no EMG findings consistent with the flail foot

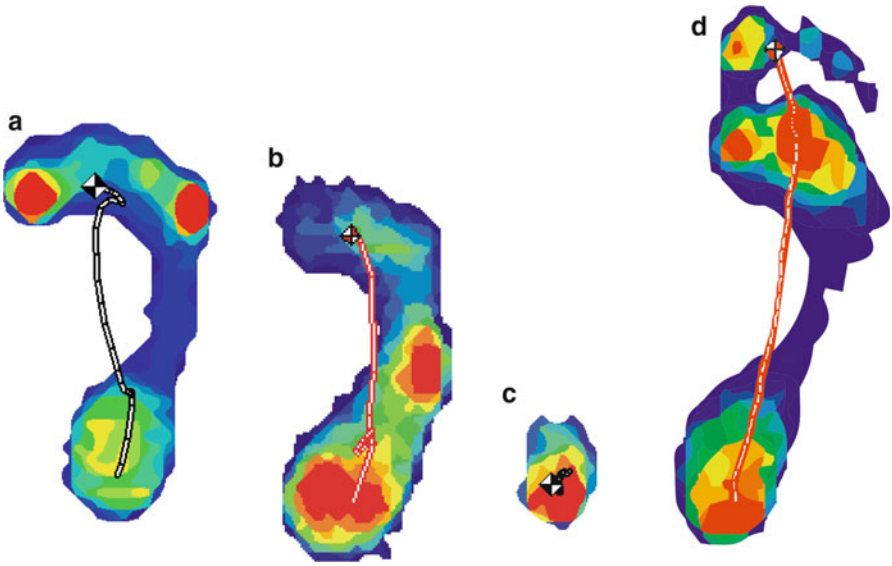

Fig. 6 Example foot pressure plots highlight common patterns found in persons with CMT including (**a**) cavus deformity with no toe contact, (**b**) increased lateral weight bearing pressure, (**c**) inability to weight bear over the distal foot in any capacity with all pressures focused under the heel in comparison to the (**d**) typically developing reference. In very young patients who cannot cooperate with a clinical exam to evaluate strength, the foot pressure plot is an excellent tool to understand plantar flexor strength

integrals have been measured with pedobarography in persons with CMT; however, their relationship to foot pain is not clear (Burns et al. 2005; Crosbie et al. 2008). As with other measurements, there is a larger variety of presentations in terms of foot pressures in persons with CMT (Fig. 6). This assessment is of particular interest in children where skin changes may not yet provide adequate information to understand the impact of cavus and adductus deformities. When used in conjunction with radiographs of the foot, the impact of anatomical abnormalities can be better understood. Foot pressures also provide an assessment of ankle plantar flexor and toe flexor strength. When toe flexor strength is compromised, the ability of the last portion of push-off is compromised, and no toe contact is made with the floor (Fig. 6a, b). When ankle plantar flexors are compromised, there is reduced ability to weight bear over the distal foot. Making this assessment by observing the foot/ankle is limited as while the flail foot may be in contact with the ground the center of pressure may not be able to move over the distal aspect of the foot (Fig. 6c). The foot pressure plot in some cases may be the only way to obtain adequate information regarding plantar flexor strength when a patient is too young or unable to understand directions in a strength assessment. Pre- versus postsurgical intervention to the foot

can also be assessed and allow for a critical examination of the impact of bony and/or soft tissue surgery outcomes during gait.

Management: Evaluation of Treatment Outcomes Using Motion Analysis

It is clear from the previous research and clinical experience that persons with CMT have a wide variety of presentations and therefore require different treatment strategies to provide optimum outcomes. Motion analysis is an excellent tool to assist in better understanding the pathomechanics with which to make more informed treatment and provide an opportunity to objectively evaluate treatment outcomes. If motion analysis is incorporated as the standard of care, it will be possible someday to provide improved care through evidenced-based medical practice. The following is a discussion of how motion analysis can be used to better define treatment and understand treatment outcomes.

Physical Therapy

Physical therapy is a common component of treatment for patients with CMT with a focus on strengthening, maintaining range of motion, and balance training with the goal of maintaining mobility. It has been shown that strength training can benefit measures of muscle strength and other outcome measures such as walking velocity (Burns et al. 2009) as well as activities of daily living in adults with CMT (Chetlin et al. 2004); however, there is limited current knowledge on how strength training impacts gait function in terms of joint kinematics and kinetics. Understanding therapy outcomes at the joint level will help to explain why some therapies may be more successful than others. For example, understanding if strengthening of the plantar flexors improved ankle sagittal plane kinematics and kinetics would help to understand why there may be benefits in walking velocity. Motion analysis could also help with targeting specific therapies to provide the most functional benefits.

Orthoses

Orthoses are very effective treatment modalities for support of the ankle when muscle weakness is a primary clinical finding. In persons with CMT, ankle instability is a very common finding resulting from weakness of the ankle plantar flexors and the medial and lateral stabilizers (Mandarakas et al. 2013). Also, clearance problems may be present as a result of weakness of the anterior tibialis and long toe extensors. Motion analysis techniques can provide insight into the impact of orthoses on ankle function that includes not only kinematic function of the ankle but also kinetic function that can explain in some cases why patients are non-compliant with their orthoses. Walking velocity has been shown to improve with the application of orthoses (Phillips et al. 2011); however, the assessment of this valuable outcome measure on its own does not provide information about the impact of the orthosis at the joint level which is required to understand why an orthosis improves walking velocity and ultimately indications for orthosis specific design. Ramdharry

(Ramdharry et al. 2012) incorporated comprehensive motion analysis and documented reduced (improved) excessive dorsiflexion in terminal stance and plantar flexion in swing with the application of an appropriately molded ankle foot orthosis (AFO). However, it is clear that in patients with CMT, one AFO design does not suit all due to the variation in presentation and degree of severity of pathology and the variety of orthosis design and purpose. Although the solid AFO design may provide excellent support for excessive ankle dorsiflexion in terminal stance due to weakness of the plantar flexors, in some cases it may overly restrict ankle movement and thus reduce the ankle power generation (Fig. 7a, b). For those persons with CMT who are more functional, this restriction in motion will have a negative impact on push-off especially during running. The posterior leaf spring orthosis (PLS) which has less support through trim lines posterior to the malleoli may result in less than adequate support in some patients but function as a solid AFO in others depending on brace stiffness, patient body weight, and plantar flexor strength which may be decreasing over time due to the disease progression.

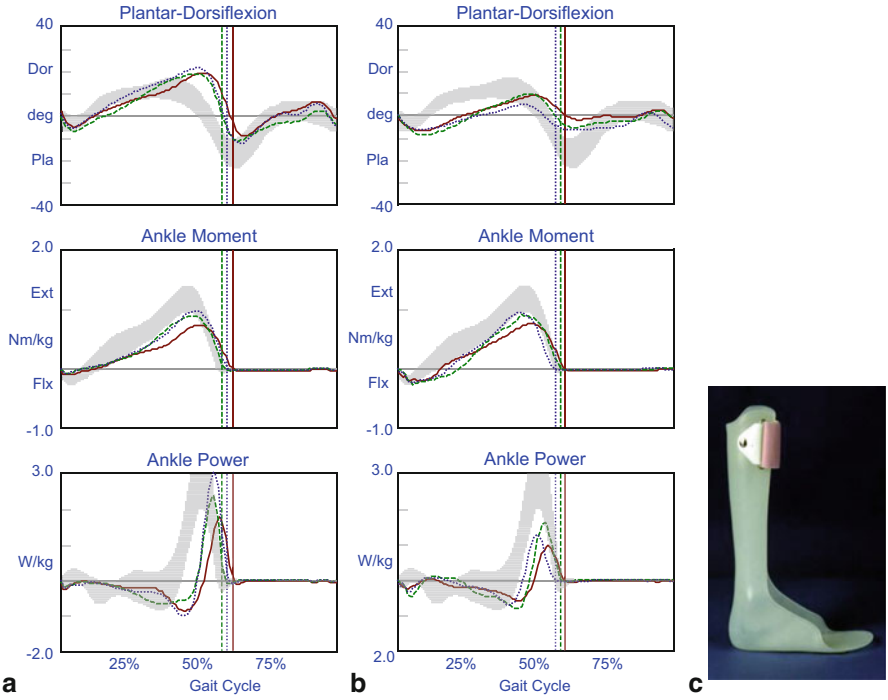

Fig. 7 Comparison of ankle sagittal plane kinematic, moment and power for (**a**) barefoot and (**b**) solid AFO walking for the *right side* in a youth with CMT. The solid AFO (**c**) can provide improved stability at the ankle in the sagittal plane by restricting excessive peak dorsiflexion in terminal stance, however, this is done at the expense peak ankle power generation which shows a decrease in the solid AFO. It is likely that this explains why some persons with CMT do not like wearing their solid AFOs especially when there are greater requirements for ankle push off such as in running

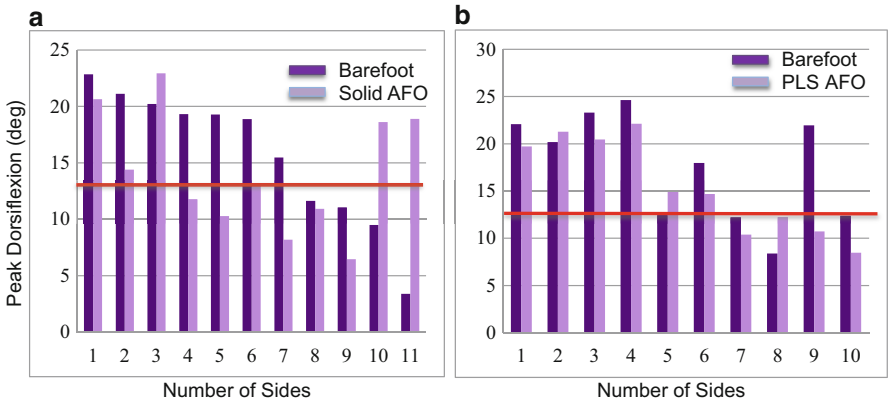

Fig. 8 Comparison peak ankle dorsiflexion during stance for barefoot versus AFO for the (**a**) solid AFO and (**b**) PLS AFO. The *red line* indicates typically developing peak dorsiflexion in stance. The large variation in values for peak dorsiflexion in terminal stance in both brace designs confirms the heterogeneous nature of CMT in terms of gait outcomes. In some cases, the brace design provided improvement and in others made no change and in other resulted in a worse outcome

We have recently reviewed AFO outcomes in children and adolescents with CMT and have shown that different AFOs (solid AFO vs. PLS), although intended to have differences in terms of function based upon their design, do not always operate as proposed. In many cases, the PLS design functions as a solid AFO and restricts most ankle motion, and in others the PLS does not provide adequate support and allows ongoing excessive ankle dorsiflexion. As for the solid AFO design, in some cases, it functions like a PLS and does not provide adequate support (Fig. 8a, b). It is also clear that hinged AFO designs, while supporting the ankle in swing and reducing excessive plantar flexion, do not provide adequate support in stance (Fig. 9a, b). Understanding orthosis function in persons with CMT using motion analysis has allowed us to clarify why some orthosis designs are more effective than others and to highlight the importance of understanding patient impairment and gait function during barefoot walking that needs to be supplemented with the AFO. Additional research is needed in this area to better match orthosis design with patient impairment. As well, there are technical considerations when using motion analysis techniques in the assessment of orthoses including marker placement and documentation of consistent ankle sagittal plane angles in both barefoot and orthosis conditions (Ounpuu 1996).

Orthopedic Surgery

Orthopedic surgery may be required when patients with CMT have foot deformity and pain that limits walking and performing activities of daily living. Surgery to address the wide variety of issues varies from individual muscle procedures (lengthenings or transfers) to complex combinations of soft tissue and bony interventions. The majority of research evaluating surgical outcomes is based upon clinical examination findings alone and, in some cases, only postoperative assessments. Motion analysis provides

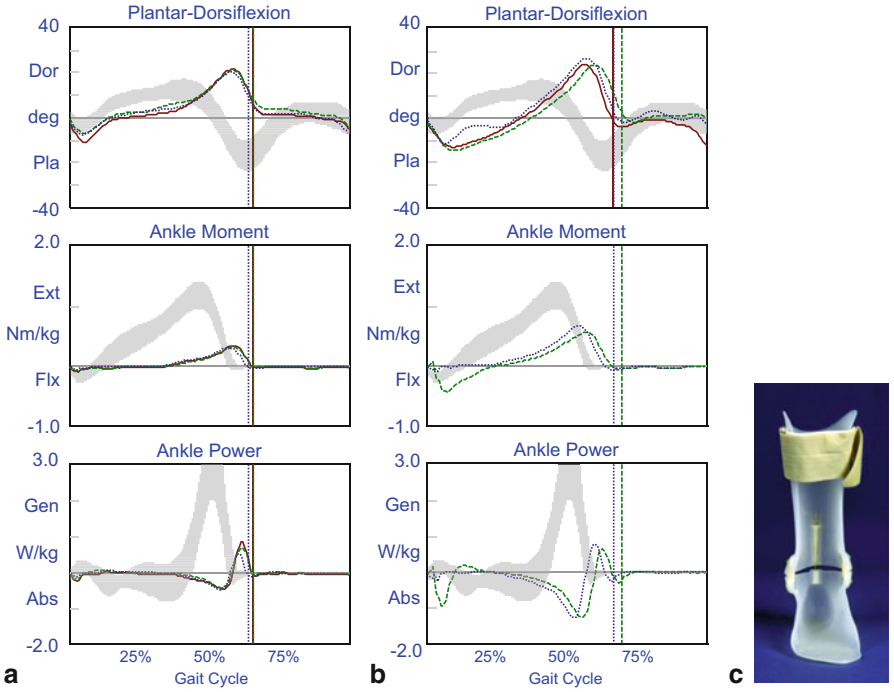

Fig. 9 Comparison of ankle sagittal plane kinematic, moment and power for (**a**) barefoot and (**b**) hinged AFO walking for the *right side* in a youth with CMT. Although the hinged AFO (**c**) can provide medial/lateral stability there is no restriction on peak ankle dorsiflexion in stance and therefore there is a continued reduction in peak ankle plantar flexor moment in stance and improvement in stability is not improved in the sagittal plane

an excellent opportunity to assist in surgical decision-making and to evaluate surgical outcomes objectively. This is possible in those clinical settings where the standard of care includes comprehensive motion analysis techniques both pre- and post-surgery. These assessments should include an evaluation of clinical impairments (muscle weakness, contracture, and bony deformity) which is integrated with gait findings documented with dynamic EMG, joint and segment kinematics, joint kinetics, and foot pressure data. This approach allows for the most informed treatment decisions and objective evaluation of outcomes when the patient has adequately recovered. This approach will allow new knowledge on a patient-by-patient basis and ultimately through research when this protocol is followed over time. Pairing treatment approaches with clear and specific indications at the level of impairment and function is needed. When interpreting postoperative data, it is important to also consider changes that might be due to the natural history of this progressive disorder.

Motion analysis has provided some insight into treatment indications from both routine clinical use and research applications, but at this time, there is only one example of comprehensive motion analysis outcomes used to assess treatment in this

patient population. It has been shown by Dreher (Dreher et al. 2014) that the transfer of the posterior tibialis tendon can reduce excessive equinus in swing which can lead to clearance issues and inappropriate prepositioning at initial contact. However, they also found reduced peak ankle plantar flexion at push-off. Joint kinetic data would provide additional insight into this finding related to plantar flexor function which is also confounded by the possibility of increased weakness over time. Orthopedic treatment outcomes have also been assessed using pedobarography with positive outcomes in terms of foot pressures; however, these measures do not correlate well with other aspects of gait (Metaxiotis et al. 2000). A study by Ward (Ward et al. 2008) evaluated 25 patients with CMT who underwent treatment for cavus foot including dorsiflexion osteotomy of the first metatarsal, transfer of the peroneus longus to the peroneus brevis, plantar fascia release, transfer of the extensor hallucis longus to the neck of the first metatarsal, and in selected cases transfer of the tibialis anterior tendon to the lateral cuneiform. Temporal and stride parameters were analyzed and revealed that those patients that had undergone the anterior tibialis tendon transfer spent less time in double-support stance phase. Additional outcomes of ankle kinematics and kinetics in the above study would have provided relevant information as to the causes of changes in temporal and stride parameters to further clarify treatment indications. This would have allowed confirmation of the surgical goals of addressing current deformity and ultimately long-term recurrence with objective measures of ankle function. Although these studies provide interesting findings, comprehensive motion analysis that includes both kinematic and kinetic outcomes would be very useful.

Individual patient cases where motion analysis has been incorporated as the standard of care can also provide important knowledge especially if this methodology is followed over the long term. This is particularly important in rare diseases when it takes time to gather a large data base of patients to study. For example, motion analysis data for an individual patient has provided some insight into the possible impact of the plantar fascia release for the correction of cavus deformity. In a patient who is in excessive equinus in stance (toe walker) and swing due to limited dorsiflexion range of motion (plantar aspect of the foot in relation to the tibia), a plantar fascia release may provide sufficient reorientation of the foot anatomically to allow for increased passive dorsiflexion range to eliminate the excessive equinus in stance and swing (Fig. 10a, b). In a patient who has normal peak ankle dorsiflexion in terminal stance but a significant cavus deformity, a plantar fascia release to correct for the cavus and varus position may provide reorientation of the foot anatomically to allow for excessive ankle passive dorsiflexion range of motion and excessive dorsiflexion in terminal stance. This may lead secondarily to increased knee flexion in stance (Fig. 11). Unmasked ankle plantar flexor weakness may in part play a role in the downside of this procedure which may still be relevant to help with foot pain. Appreciating the complex relationship between maximum dorsiflexion range of motion, plantar flexor strength, patient body weight, and the extent of cavus deformity is all more possible with motion analysis. Learning from these individual cases is an important step toward the goal of improving treatment outcomes.

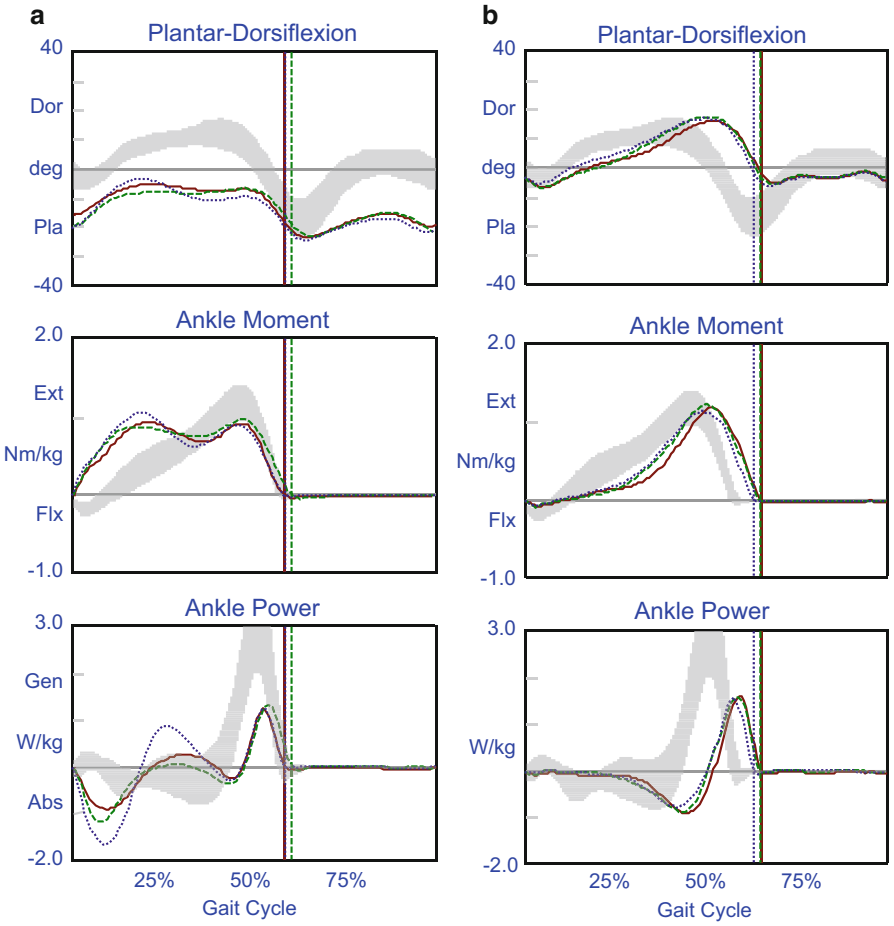

Fig. 10 Comparison of the sagittal plane ankle kinematics, moments and powers for (**a**) pre and (**b**) several years post orthopaedic lengthening of the plantar fascia in an child with CMT. Three gait cycles on the *right side* are plotted for each condition. Improvements in ankle sagittal plane motion in stance and swing and ankle moment modulation due to a heel initial contact without reduction in ankle power generation are noted. Objective documentation of the outcome of this procedure using motion analysis can help to clarify surgical indications

Future Directions

The application of comprehensive motion analysis techniques for clinical evaluation as part of the standard of care and in research settings for persons with CMT is relatively new compared to other complex gait pathologies such as cerebral palsy. As a result there is still much to learn and new developments needed to optimize motion measurement for this pathology. Future effort should focus on the following:

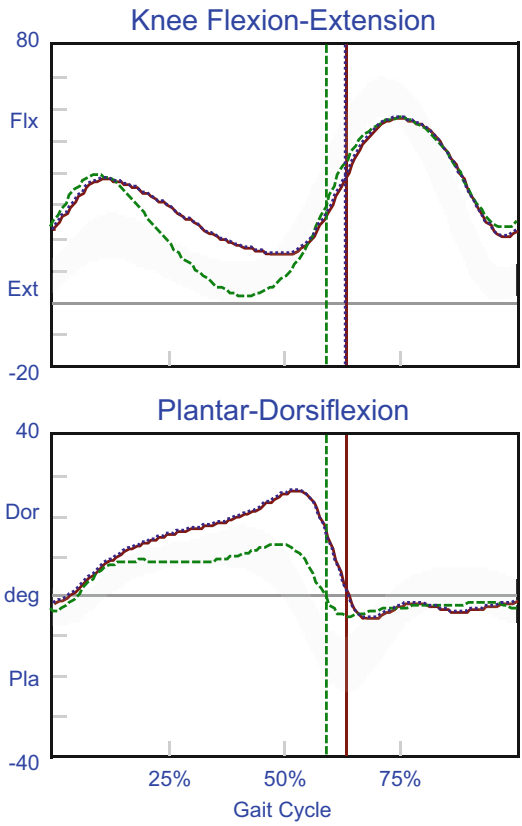

Fig. 11 Comparison of the knee and ankle sagittal plane kinematics for pre (*dashed*) versus post (*solid*) orthopaedic surgical outcome for plantar fascia release. Increased peak ankle dorsiflexion and associated knee flexion may be due to multiple causes: (**a**) ongoing disease process of increasing weakness of the plantar flexors over time and/or (**b**) unmasking of existing ankle plantar flexor weakness through increased dorsiflexion range of motion

(a) Application of a more comprehensive foot model is needed to better understand the complex relationship between cavus deformity, plantar flexor strength, and passive ankle dorsiflexion range of motion. The foot model needs to include measurement of the extent of cavus and may need to incorporate radiographic information to allow for the most accurate assessment. Understanding how the extent of cavus deformity impacts available plantar flexor length and masks weakness is critical for treatment decision-making at the ankle/foot in these patients.

(b) The presentation of CMT is heterogeneous with varying clinical findings, severity, as well as disease progression. Establishing if there is any link between impairments and associated gait function with phenotype will provide a better ability to determine prognosis for future function at the time of diagnosis and lead to treatment guidelines based upon phenotype.

(c) CMT is a progressive disease that typically results in increasing weakness and associated gait issues over time. Disease progression, however, is patient dependent and not a lot is known about expectations for decline for a given individual with CMT. Long-term natural progression studies are needed which hopefully can be linked eventually to phenotype. Achieving this goal is difficult as disease progression in many is not rapid so long-term studies are needed.

(d) Treatment of gait issues for this patient population focuses on improving or maintaining gait function and reducing foot/ankle pain. There is very limited objective documentation of orthopedic and other treatment outcomes such as bracing in terms of comprehensive motion analysis. Systematic reviews of surgical outcomes are needed to assist in identifying specific treatment indications and expectations. This research is further complicated by disease progression which needs to be taken into account.

(e) Finally, establishing a diagnosis of CMT is difficult, and in the initial phases, it is often confused with other diagnoses such as idiopathic toe walking to cavo-varus deformity. Initial steps include a detailed clinical assessment and should also include a family history. If there is a suspected CMT diagnosis, often genetic testing and nerve conduction tests are recommended to confirm a diagnosis and phenotype. These later tests are often not completed due to expense, and nerve conduction tests can be painful. Therefore, establishing functional biomarkers related to movement would be helpful in the initial stages of this disease to help correctly diagnosis and therefore treat appropriately common initial findings of cavo-varus foot deformity in some and toe walking in others. These biomarkers may include any or a combination of the following: ankle kinematic or kinetic variables, EMG signal analysis, and muscle impedance assessments. Additional investigation is needed to determine if these or other biomarkers can be established.

Cross-References

- ▶ 3D Dynamic Pose Estimation from Marker-Based Optical Data
- ▶ Assessing Pediatric Foot Deformities by Pedobarography
- ▶ Functional Effects of Foot Orthoses
- ▶ Interpreting Joint Moments and Powers in Gait
- ▶ Kinematic Foot Models for Instrumented Gait Analysis

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