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# Charcot-Marie-Tooth Disease of the Foot and Ankle: Imaging Features and Pathophysiology

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Charcot-Marie-Tooth disease (CMT) is the most common inherited peripheral polyneuropathy, resulting in length-dependent motor and sensory deficiencies. Asymmetric nerve involvement in the lower extremities creates a muscle imbalance, which manifests as a characteristic cavovarus deformity of the foot and ankle. This deformity is widely considered to be the most debilitating symptom of the disease, causing the patient to feel unstable and limiting mobility. Foot and ankle imaging in patients with CMT is critical for evaluation and treatment, as there is a wide range of phenotypic variation. Both radiography and weight-bearing CT should be used for assessment of this complex rotational deformity. Multimodality imaging including MRI and US is also important to help identify changes in the peripheral nerves, diagnose complications of abnormal alignment, and evaluate patients in the perioperative setting. The cavovarus foot is susceptible to distinctive pathologic conditions including soft-tissue calluses and ulceration, fractures of the fifth metatarsal, peroneal tendinopathy, and accelerated arthrosis of the tibiotalar joint. An externally applied brace can assist with balance and distribution of weight but may be appropriate for only a subset of patients. Many patients will require surgical correction, which may include soft-tissue releases, tendon transfers, osteotomies, and arthrodesis when necessary, with the goal of creating a more stable plantigrade foot. The authors focus on the cavovarus deformity of CMT. However, much of the information discussed may also be applied to a similar deformity that may result from idiopathic causes or other neuromuscular conditions.

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**Abbreviation:** CMT = Charcot-Margot-Tooth disease

## TEACHING POINTS

- The characteristic progressive pes cavovarus deformity of CMT occurs secondary to the imbalance created by asymmetric muscle involvement in the lower extremities.
- Owing to the complexity and multidimensionality of the cavovarus deformity in CMT, weight-bearing CT plays an important role in assessing hindfoot alignment.
- Patients with CMT may demonstrate normal calcaneal pitch angles, as the CMT cavovarus deformity is primarily forefoot driven owing to plantar flexion of the first ray. An elevated calcaneal pitch angle is more commonly seen in patients with idiopathic pes cavus.
- Patients with CMT are highly susceptible to fractures of the fifth metatarsal owing to the extreme mechanical overload on the lateral border of the foot.
- The goal of surgery is to create a more stable plantigrade foot to preserve mobility and minimize bracing. To accomplish this, the surgeon must balance muscle pull by using tendon transfers, release soft-tissue contractures, and realign the weight-bearing planes of the foot with wedge osteotomies.

## Introduction

Charcot-Marie-Tooth disease (CMT) is a group of genotypically diverse conditions that manifest as a length-dependent peripheral polyneuropathy that leads to atrophy and weakness of the distal muscles, decreased peripheral sensation, and reduced tendon reflexes. It is the most common hereditary motor and sensory neuropathy, with an estimated worldwide prevalence of 10–28 persons per 100 000 (1). Ongoing developments in next-generation sequencing have helped identify over 80 associated genes and several different pathophysiologic pathways, which help to explain the significant phenotypic variability of the disorder (1). Despite this variability, the vast majority of patients with CMT will develop progressive cavovarus deformity of the foot and ankle due to asymmetric muscle involvement in the lower extremities, which may be debilitating in severe cases. Gradually, the deformity leads to early-onset arthritis because of the inability to normally distribute pressure in the joints (2–7). An understanding of the disease and its cardinal manifestation allows the radiologist to potentially assist in diagnosis, identify common associated pathologic conditions, and accurately recognize postoperative imaging appearances and complications in patients who have undergone surgical correction.

Although CVS is the most common cause, it is not the sole cause of cavovarus foot deformity (8). Other causes include infection such as poliomyelitis; other neurologic disorders including cerebral palsy, spinal dysraphism, and muscular dystrophy; acquired pathologic conditions including stroke, spinal cord tumor, or trauma; and idiopathic causes (6,8–10). In patients presenting with cavovarus foot, one distinction to make is whether the cavus deformity is hindfoot driven or forefoot driven. Hindfoot-driven pes cavus results in a vertically oriented calcaneus and can be due to a weakened gastrocnemius-soleus complex, which may be seen with poliomyelitis and various spinal patho-

logic conditions (9). Idiopathic pes cavus also tends to be hindfoot driven (11). However, patients with CMT usually present with a forefoot-driven pes cavovarus deformity. The pathophysiology of this deformity is complex, although a primary driver involves plantar flexion of the first ray. Despite this distinction, the cause of a cavovarus foot deformity may be unclear on the basis of imaging findings alone, and diagnosis will depend on a thorough clinical history and physical examination. In this article, we focus on the forefoot-driven cavovarus deformity seen in CMT, but many concepts discussed here can also be applied to the general cavus foot.

## Background

CMT, formerly referred to as “peroneal muscular atrophy,” was originally described in France in 1886 by Jean-Martin Charcot and Pierre Marie, who incorrectly characterized the disease as a myelopathy, potentially the familial form of spinal muscular atrophy (12). Shortly after this, Howard Henry Tooth independently described the condition in England. Tooth correctly identified CMT as a peripheral neuropathy by noting the occasional occurrence of sensory features (12). One of the largest descriptions of a cohort with the disease was published in 1952 by England and Denny-Brown (12), who studied an extended family of 303 members, all with varying forms of the disease ranging from mild to severe. Those with the mildest form demonstrated slight pes cavus, while patients with advanced disease had extensive sensory and trophic changes. Some distinguishing features from their study include a wide highly arched foot, toes that point upward and then curl sharply downward, near-complete muscle atrophy below the knee with the peroneal muscles always involved, and loss of sensation in severe cases leading to infection (12). These features remain the hallmark of disease manifestation today.

## Pathophysiology

CMT is a group of hereditary conditions, all of which may be passed down with varying inheritance patterns. The rate of de novo genetic mutations in the most common subtype of CMT, CMT type 1A (CMT1A), is reportedly as high as 19% (13), making family history an important but not exclusive feature of the disease. A case report of two sets of homozygous male twins reveals variability in disease severity, with differing ages of onset and functional weakness (14). These findings highlight the importance of environmental modulation and endogenous factors in regulating gene expression in CMT.

CMT is classified into subtypes on the basis of the affected gene, with over 80 associated genes identified at this time. However, roughly 90% of patients with CMT confirmed by genetic testing results have mutations in one of a few genes (1,15). These genes include peripheral myelin protein-22 (*PMP22*), gap junction  $\beta$ -1 (*GJB1*), myelin protein zero (*MPZ*), and mitofusin-2 (*MFN2*) or ganglioside-induced differentiation-associated protein 1 (*GDAP1*), depending on geographic location. *MFN2* is more common in North America and Northern Europe, while *GDAP1* is more widespread in the Mediterranean region (1). Table 1 outlines the primary CMT subtypes and their corresponding genes, inheritance patterns, and frequencies. Next-generation sequencing

**Table 1: Primary Subtypes of CMT**

Subtype	Genetic Mutation	Inheritance Pattern	Frequency (%)*	Motor Conduction Velocity
CMT1A	<i>PMP22</i> duplication	Autosomal dominant	60.5	Demyelinating
CMTX1	<i>GJB1</i> mutation, most commonly loss of function	X-linked dominant	16.7	Intermediate
CMT1B	<i>MPZ</i> frameshift mutation	Autosomal dominant	9.4	Demyelinating
CMT2A	<i>MFN2</i> mutation	Autosomal dominant	4.4	Axonal

Source.—Reference 15.

\* Frequency of occurrence in genetically confirmed cases of CMT.

technology continues to help identify novel genes, adding complexity to the current classification system.

### Clinical Presentation

The typical order of muscle involvement in CMT is characteristic, beginning with the intrinsic foot musculature, then the extensor compartment, followed by the peroneus brevis. In highly advanced cases, the calf muscles may also be affected. In regard to the extensor compartment, the tibialis anterior muscle is always involved, whereas there is variable involvement of the extensor hallucis longus and extensor digitorum longus. While there is a clear length-dependent pattern, there is only speculation about why certain muscles are spared, most notably the posterior tibialis, peroneus longus, and, in some cases, long toe extensors (extensor hallucis longus or extensor digitorum longus). One theory hypothesizes that an element of compression neuropathy is in play and notes the similarities between muscle involvement in CMT and the disease entity known as hereditary neuropathy with liability to pressure palsies (6). Interestingly, both CMT1A and hereditary neuropathy with liability to pressure palsies display abnormalities in the expression of *PMP22*, which links the two conditions (6). The majority of patients with CMT will demonstrate similar severity of muscle involvement in both limbs, but up to 5% may display significant asymmetry, a finding associated with more advanced disability (16).

The characteristic progressive pes cavovarus deformity of CMT occurs secondary to the imbalance created by asymmetric muscle involvement in the lower extremities. A strong posterior tibialis muscle overpowers the weakened peroneal musculature (the peroneus brevis muscle is weaker than the peroneus longus muscle), leading to hindfoot varus or midline deviation of the calcaneus with respect to the tibia. The severely weakened anterior tibialis muscle, which normally functions to dorsiflex the ankle, is supplemented by the extensor digitorum longus and extensor hallucis longus. The latter tendons insert on the digits, and their continual overuse as secondary ankle dorsiflexors results in extension at the metatarsophalangeal joints. Hyperextension at the metatarsophalangeal joints then tightens the plantar fascia, which pulls the calcaneus and metatarsal heads closer to one another and heightens the longitudinal arch, contributing to pes cavus (6,9,17). A relatively preserved peroneus longus acts to disproportionately plantar flex the first ray, leading to midfoot supination or inversion of the midfoot along the axis of the cuneiforms (6,7). Weakening and contracture of the intrinsic

foot musculature lead to flexion at the proximal and distal interphalangeal joints and unopposed extension at the metatarsophalangeal joints, resulting in claw toe deformities (6,7). Table 2 and Figure 1 highlight these characteristic features with clinical and radiographic correlation.

The most common presenting concerns for patients with CMT include poorly fitting shoes, frequent falls, recurrent ankle sprains, foot pain, and cramping. Physical examination reveals cavovarus foot deformity, reduction of lower limb reflexes, the typical pattern of lower extremity muscle atrophy and weakness, impaired ankle range of motion, and altered gait (3,6,18,19). In some patients, the intrinsic muscles of the hands may be involved, leading to atrophy and weakness with impaired abduction of the digits (6,7). In patients who present with bilateral cavovarus feet, the reported prevalence of CMT is as high as 79%, increasing to 91% if there is a family history of the disease (18). Diagnostic workup, in addition to history, physical examination, and imaging evaluation, may include genetic testing, as nerve conduction velocity and electromyography studies can be equivocal (18).

### Imaging Features

#### Radiography

Radiography is the mainstay for imaging evaluation of CMT skeletal manifestations in the lower extremity. For all patients with or suspected to have the disease, the orthopedic surgeon should, at a minimum, obtain standing anteroposterior, mortise or oblique, and lateral views of the bilateral ankles, as well as standing anteroposterior, oblique, and lateral views of the bilateral feet. Specialists also recommend obtaining hindfoot alignment views (20).

There are numerous angles and measurements that can assist in the radiographic evaluation of a patient with CMT. For the sake of simplicity, a few key measurements are highlighted here. Table 2 covers the method for quantification as well as each measurement's contribution to the CMT deformity. An important angle to recognize is the angle between the long axis of the talus and the long axis of the first metatarsal on a lateral view, also known as the Meary angle (7,11). The normal lateral talus–first metatarsal angle is close to zero or minimally dorsiflexed (up to 5°). Greater than 5° of plantar flexion is abnormal and should raise suspicion for an underlying abnormality, resulting in forefoot-driven cavus. (6,7) Additional radiographic measurements include calcaneal pitch, calcaneal moment arm, naviculocuboid overlap,

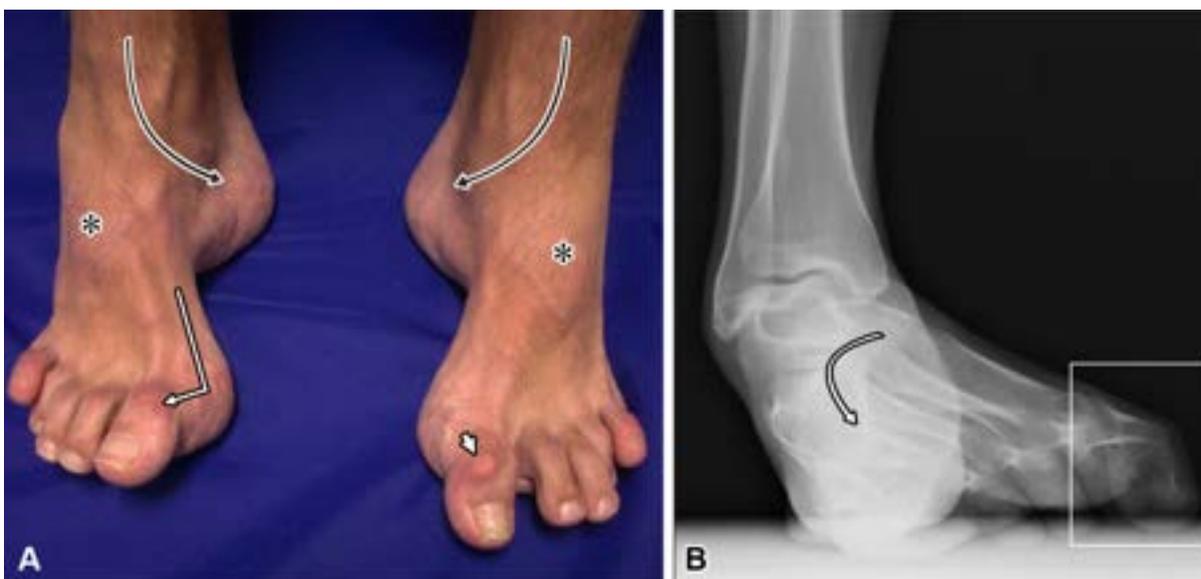
**Table 2: Angle Measurements in Cavovarus Deformity**

Deformity	Definition	Measurement	View	Reference Lines	Pertinent Values
Pes cavus	Multiplane deformity describing a high arched foot with increased plantar concavity	Calcaneal pitch angle (may be normal in CMT)	Lateral foot	Angle between plantar soft tissues and inferior margins of the calcaneus	Normal: 17°–20° >30° hindfoot cavus
Hindfoot varus	Midline tilt of the calcaneus relative to the tibia	Calcaneal apparent moment arm	Hindfoot alignment view	Perpendicular distance between longitudinal midaxis of the tibia and lowest point of the calcaneus	Normal: <8 mm Borderline: 8–15 mm >15-mm hindfoot varus
Midfoot supination	Inversion of midfoot	Naviculo-cuboid overlap	Lateral foot	CC length of naviculo-cuboid overlap divided by CC length of cuboid	<0.4 midfoot supination
Forefoot adduction	Medial deviation of the navicular (ie, overcoverage of medial talar head)	Talonavicular coverage angle	AP foot	Angle between the line connecting the whole talar head articular surface with the line connecting the contact surface between the talus and navicular	Reported mean of 12° in patients with CMT (24) Normal: neutral or slight lateral deviation of the navicular
		AP talus–first MT angle	AP foot	Angle between long axis of talus and long axis of first MT	Reported means for CMT range from 14°–33° (11,23) Normal: 7° ± 5°
Forefoot valgus	Plantar flexion of the first MT resulting in eversion of forefoot	Lateral talus–first MT angle*	Lateral foot	Angle between long axis of the talus and long axis of the first MT	Normal: 0°–5° dorsiflexion >5° plantar flexion indicates forefoot cavus

Sources.—References 8, 10, 19–22, 28.

Note.—AP = anteroposterior, CC = craniocaudal, MT = metatarsal.

\* Meary angle.



**Figure 1.** Characteristic features of the cavovarus foot in a 17-year-old adolescent boy with CMT. (A) Clinical photograph demonstrates bilateral hindfoot varus alignment (curved arrows), heightening of the longitudinal arches (\*), plantar flexion of the first ray with hyperextension of the first metatarsophalangeal joint (right angle arrow), and ulceration over the left first interphalangeal joint (short white arrow), which is hyperflexed. (B) Oblique radiograph of the right ankle shows midfoot supination, or inversion of the midfoot along the cuneiforms (arrow), and claw toe deformity of the first digit with flexion at the first interphalangeal joint (rectangle).



**Figure 2.** Key radiographic measurements and signs for evaluation of CMT.

(A, B) Lateral radiographs of the foot in a 25-year-old woman with CMT (A) and a 22-year-old healthy female control patient (B) show the Meary angle, or the lateral talus–first metatarsal angle (black angle). The findings in the patient with CMT (A) indicate plantar flexion, while the angle in the control patient (B) approaches 0. The calcaneal pitch (white angle) is normal in both patients. The measurements in the patient with CMT indicate a naviculocuboid overlap of 0 (arrow in A), while the control patient has a naviculocuboid overlap measurement of roughly 0.5 (dashed circle in B). The image of the patient with CMT shows the double talar dome sign (dashed rectangle in A) and the sinus tarsi see-through sign (dashed circle in A).

(C, D) Anteroposterior radiographs of the foot in the same two patients as in A and B show the anteroposterior talus–first metatarsal angle (black angle), which is significantly larger in the patient with CMT (C) versus in the control patient (D). The talonavicular coverage angle (white angle) indicates overcoverage of the medial talar head in the patient with CMT (C) and slight lateral deviation of the navicular bone in the control patient (D). (E) Radiograph of the hindfoot in a 53-year-old woman with CMT shows calculation of the calcaneal apparent moment arm with a line drawn through the long axis of the tibia (black line) and measurement of the distance to the lowest part of the calcaneus (white line). It is increased in this patient and is compatible with hindfoot varus.



(C, D) Anteroposterior radiographs of the foot in the same two patients as in A and B show the anteroposterior talus–first metatarsal angle (black angle), which is significantly larger in the patient with CMT (C) versus in the control patient (D). The talonavicular coverage angle (white angle) indicates overcoverage of the medial talar head in the patient with CMT (C) and slight lateral deviation of the navicular bone in the control patient (D). (E) Radiograph of the hindfoot in a 53-year-old woman with CMT shows calculation of the calcaneal apparent moment arm with a line drawn through the long axis of the tibia (black line) and measurement of the distance to the lowest part of the calcaneus (white line). It is increased in this patient and is compatible with hindfoot varus.

talonavicular coverage angle, and anteroposterior talus–first metatarsal angle (8,10,11,21–25). These measurements are illustrated in Figure 2.

There are a few radiographic signs that can be useful when interpreting foot and ankle radiographs, including the sinus tarsi “see-through” sign and the double talar dome sign (Fig 2). Both of these signs are a function of hindfoot external rotation, which occurs in CMT secondary to hindfoot varus in conjunction with forefoot adduction (26). Abnormal bony morphology in patients with CMT, namely decreased sagittal talar neck declination and a more horizontally oriented posterior calcaneal facet, may also contribute to hindfoot external rotation (27). This alignment of the hindfoot allows the long axis of the sinus tarsi to be parallel with the x-ray beam directed toward the medially placed cassette, thus making the sinus tarsi see through. Similarly, this external rotation allows simultaneous visualization of both the medial and lateral aspects of the talar dome, creating a double dome. Metatarsal stacking is a result of midfoot supination, or inversion of the midfoot through the axis of the cuneiforms (Fig 3).

### Computed Tomography

Owing to the complexity and multidimensionality of the cavovarus deformity in CMT, weight-bearing CT plays an important role in assessing hindfoot alignment. Cone-beam weight-bearing CT acquires multiplanar images under physiologic load in the standing position with decreased radiation when compared with standard fan-beam CT (28). Specialized software can then be used to reliably calculate specific two- and three-dimensional measurements, which may be difficult to accomplish with traditional radiography owing to increased superimposition with complex deformities (11,21). In some cases, the software is fully automated, eliminating the concern for inter- and intraobserver reliability (24,27).

One group of investigators (11) used weight-bearing CT to study the cavovarus deformity in patients with CMT and patients with idiopathic pes cavus compared with healthy control patients. They found that while the height of the medial longitudinal arch and degree of forefoot adduction were similar between the two groups with cavovarus deformity, patients with CMT demonstrated increased midfoot supination and worsened hindfoot alignment, implying



**Figure 3.** Multidimensional multimodality preoperative imaging of the foot in a 7-year-old girl with advanced CMT. Three-dimensional weight-bearing CT image (A), coronal CT reformation through the hindfoot (B), axial CT reformation through the midfoot at the level of the metatarsal bases (C), and oblique radiograph (D) demonstrate the classic features of the deformity, including hindfoot varus or midline tilt of the calcaneus with respect to the tibia (white lines in B and D), metatarsal stacking (double-headed arrow in C), and severe lateral foot weight bearing, resulting in chronic cortical thickening of the fifth metatarsal (circle in C and D).

more severe rotational anomalies. Additionally, the authors discovered that patients with CMT often demonstrated paradoxically normal calcaneal pitch angles secondary to severe midfoot supination, which overwhelmed the dorsiflexion of the calcaneus (11). Patients with CMT may demonstrate normal calcaneal pitch angles, as the CMT cavovarus deformity is primarily forefoot driven owing to plantar flexion of the first ray. An elevated calcaneal pitch angle is more commonly seen in patients with idiopathic pes cavus. (6,7,11)

The authors of another recent study (24) used weight-bearing CT to look at the rotational difference in coronal and axial planes in patients with CMT versus healthy control patients to assess the contribution of each tarsal joint to the multiplanar deformity of CMT. Findings indicated that the peak coronal rotational deformity in CMT occurred at the transverse tarsal joints (talonavicular and calcaneocuboid), and the peak adduction deformity in the axial plane occurred at the talonavicular joint, resulting in overcoverage of the medial talar head (24).

Weight-bearing CT can be used to create multidimensional reformations, more easily illustrating the complex rotational malalignment of the cavovarus deformity in patients with CMT. Figure 3 highlights an example of a three-dimensional reformation with associated two-dimensional CT images and a radiograph in a 7-year-old girl with CMT. These three-dimensional images are used by the surgeons at our institution for preoperative planning and intraoperative decision making. Weight-bearing CT has also reportedly been used to create three-dimensional printed models for investigation of different calcaneal osteotomy techniques in correcting varus hindfoot deformity in patients with CMT (29,30).

### MRI and US

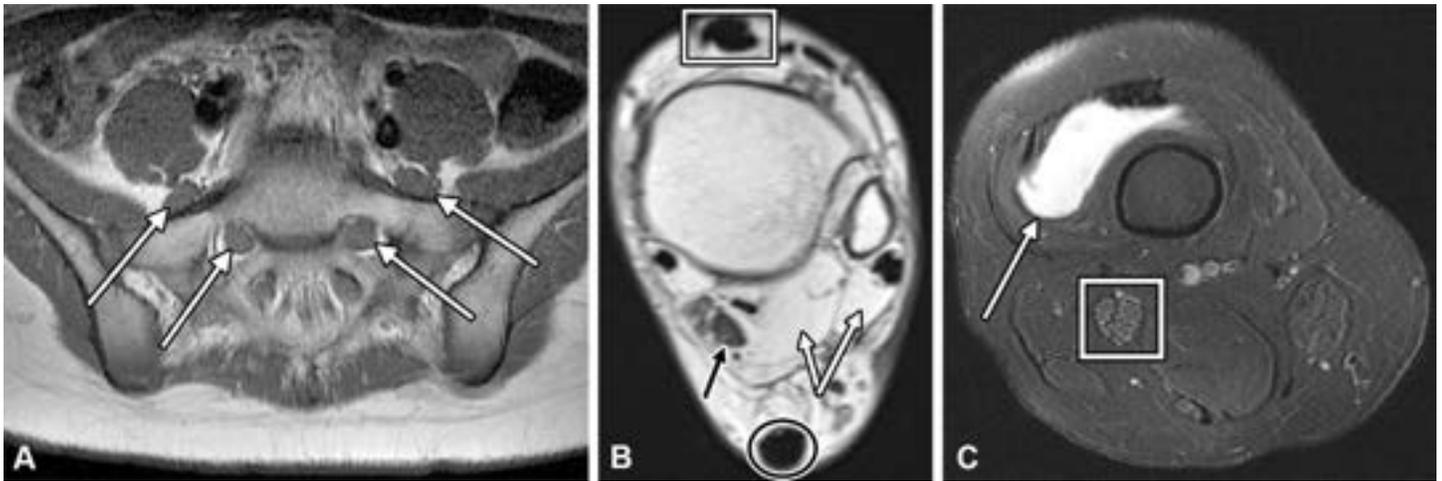
A primary role of MRI in the evaluation of patients with CMT is to help identify potential soft-tissue pathologic conditions associated with the cavovarus deformity. There is currently no

clinical role for MRI in quantifying the degree of muscle involvement in these patients. Theoretically, MRI should be able to help detect the earliest manifestation of the disease, namely increased water content in the involved muscles. In the setting of chronic disease, MR images can illustrate the accumulation of intramuscular fat. These findings, however, can be difficult to quantify, especially when looking for subtle changes. Researchers have investigated specific MRI measurements, including fat fraction and magnetization transfer ratio, to quantify the response to novel therapies (31). Furthermore, the authors (31) found that fat fraction data correlated more highly with strength and function when compared with clinical examination and myometric measures. Fat fraction analysis has also been used to quantify the fat content of peripheral nerves, which has been shown to be higher in patients with CMT (32).

MRI and US can both be used to quantify and study the cross-sectional area of the peripheral nerves in patients with CMT. Studies show an increase in nerve diameter in patients with CMT when compared with age-matched control patients (32,33). In addition, the cross-sectional diameter disproportionately increases with age in patients with CMT compared with healthy patients, suggesting ongoing nerve changes during childhood. Unsurprisingly, the nerve cross-sectional diameter was found to correlate with disease severity (33). It has been suggested that there may be a role for US to play in the early diagnosis of patients with CMT. Figure 4 illustrates the MRI appearance of the peripheral nerves in three patients with CMT.

### Complications of Abnormal Alignment

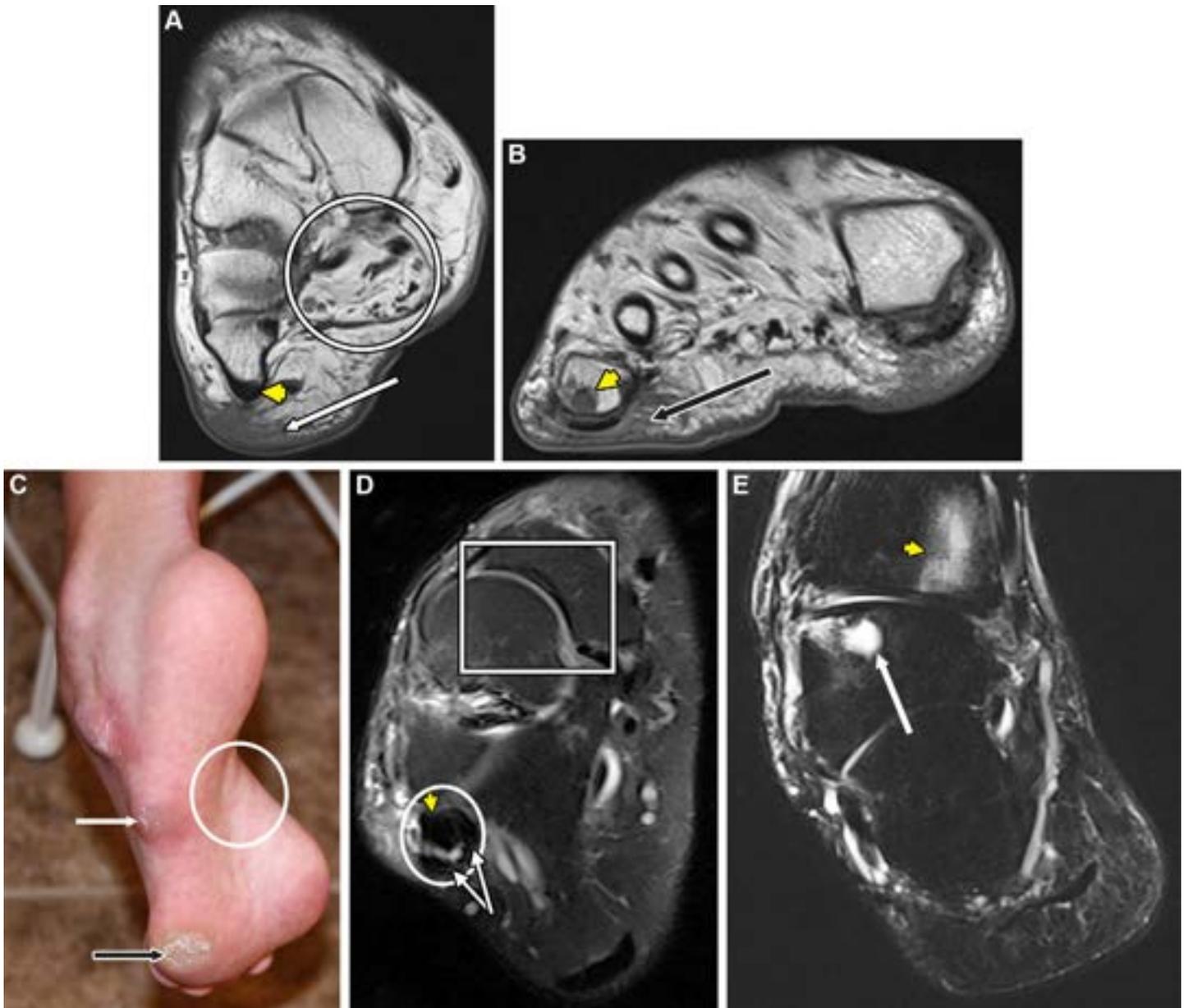
The cavovarus deformity of CMT lends itself to characteristic pathologic conditions (Figs 5, 6). Notably, fifth metatarsal fractures have been shown to be associated with varus heel alignment (34). Patients with CMT are highly susceptible to fractures of the fifth metatarsal owing to the extreme mechanical overload on the lateral border of the foot. These fractures, including stress fractures, can occur anywhere on the fifth



**Figure 4.** MRI appearance of peripheral nerves in three patients with CMT. (A) Axial T1-weighted MR image through the upper sacrum in an 11-year-old girl shows significant enlargement of the lumbosacral nerve roots (arrows). (B) Axial T1-weighted MR image through the ankle in a 40-year-old woman shows moderate enlargement of the posterior tibial nerve (black arrow). The patient had undergone posterior tibial tendon transfer, which can be seen crossing over the tibialis anterior tendon dorsally (rectangle). There is complete fatty replacement of the flexor musculature (white arrows) and mild Achilles tendinosis (circle). (C) Axial T2-weighted fat-saturated MR image through the distal femur in a 53-year-old woman shows marked hypertrophy of the sciatic nerve (square). The patient also has a moderate knee effusion (arrow) secondary to severe tricompartmental arthrosis of the knee (not shown).



**Figure 5.** Complications of abnormal alignment in four patients with CMT. (A) Lateral radiograph of the foot in a 20-year-old woman demonstrates a nondisplaced fracture of the proximal fifth metatarsal (black circle). Note the radiographic sinus tarsi see-through sign (white dashed circle) and the double talar dome sign (dashed rectangle) indicative of cavovarus alignment. (B) Anteroposterior radiograph of the foot in a 42-year-old woman shows a chronic fracture deformity of the fifth metatarsal neck (circle) and metatarsal stacking (double-headed arrow). (C) Oblique radiograph of the foot in a 36-year-old man shows a fracture of the proximal fifth metatarsal necessitating screw fixation (rectangle). This patient has also undergone first metatarsal osteotomy (dashed circle) and calcaneal osteotomy (arrow). (D) Sagittal CT image in a 59-year-old man shows secondary ossification of the Achilles tendon from long-standing contracture (rectangle).



**Figure 6.** Complications of abnormal alignment in a 37-year-old man with CMT. (A) Short-axis T1-weighted MR image through the midfoot at the level of the fifth metatarsal base shows fifth metatarsal cortical thickening (yellow arrow), chronic pressure-related changes within the subcutaneous fat plantar to the fifth metatarsal base (white arrow), and atrophy of the intrinsic foot musculature (circle). (B) Short-axis T1-weighted MR image through the forefoot at the level of the fifth metatarsal head shows a degenerative subcortical cystlike change within the fifth metatarsal (yellow arrow) and underlying chronic pressure-related changes within the subcutaneous fat (black arrow). (C) Clinical photograph of the undersurface of the foot shows the corresponding soft-tissue calluses at the level of the fifth metatarsal base (white arrow) and head (black arrow) and atrophy of the foot muscles (circle). (D) Axial T2-weighted fat-saturated MR image of the ankle at the level of the talar head shows forefoot adduction (ie, overcoverage of the medial talar head; rectangle), a high-grade split tear of the peroneus brevis (white arrows), peroneus longus tendinosis (yellow arrow), and peroneal tenosynovitis (circle). (E) Coronal short- $\tau$  inversion-recovery MR image through the ankle shows advanced tibiotalar arthrosis with narrowing of the medial tibiotalar joint space and degenerative subchondral bone marrow edema and cystlike change within the lateral talar dome (white arrow) and medial tibial plafond (yellow arrow).

metatarsal and may necessitate screw fixation if complicated by nonunion (35). Other findings related to lateral foot overload include cortical thickening of the fifth metatarsal secondary to chronic stress response, reactive subchondral bone marrow signal intensity changes in the fifth metatarsal, and soft-tissue callus formation at the fifth metatarsal base and head.

Soft-tissue ulcers may also form over the interphalangeal joints owing to claw toe deformities and friction from overlying shoes. Hyperextension at the metatarsophalangeal

joints and claw toes leads to increased pressure on the metatarsal heads, which may cause soft-tissue changes including ulceration, possibly resulting in infection and osteomyelitis at advanced stages. Soft-tissue contractures, such as of the Achilles tendon and plantar fascia, may manifest as secondary ossification. Hindfoot varus alignment leads to accelerated arthrosis at the tibiotalar joint and pathologic conditions of the peroneal tendons, including tendon tears and tenosynovitis (25).

**Table 3: Surgical Correction of CMT Cavovarus Foot**

Procedure	Technique	Purpose
Medial soft-tissue releases	Release medial talonavicular joint capsule, spring ligament, and rarely subtalar joint capsule	Relieve contractures to derotate midfoot supination into a plantigrade foot
PTT release and transfer	Through interosseous membrane to dorsum of foot, secured to the middle or lateral cuneiform	Supplement anterior tibialis to augment ankle dorsiflexion; remove deforming force of PTT
EHL and EDL transfers	From digits to metatarsals or cuneiforms	Remove deforming force resulting in claw toes deformities; increase power as secondary ankle dorsiflexors
IP arthrodesis	Screw fixation across the IP joint	May be performed to counteract unopposed flexion at IP joints after EHL or EDL transfer
Peroneal tendon transfer	Pulvertaft weave of the peroneus brevis into the peroneus longus distal to the fibula	Strong PL augments weak PB with eversion; remove deforming plantar flexion force on first ray
Plantar fascia release	Medial incision centered over the insertion on the posterior calcaneal tuberosity	Often contracted in patients with CMT
Calcaneal osteotomy	Closing wedge lateralizing osteotomy in conjunction with rotation of the tuberosity	Lateralize weight-bearing axis of calcaneus and pull of Achilles
First (and possibly second) MT osteotomy	Dorsal closing wedge osteotomy of the base	Elevate and realign the weight-bearing plane of MT heads

Sources.—References 5, 16, 19, 29, and 30.

Note.—EDL = extensor digitorum longus, EHL = extensor hallucis longus, IP = interphalangeal, MT = metatarsal, PB = peroneus brevis, PL = peroneus longus, PTT = posterior tibial tendon,

## Treatment

### Conservative

Treatment options vary on the basis of the severity of the foot deformity. The first-line treatment in patients presenting primarily with weakness and a mild deformity is ground-reaction ankle foot orthoses (20). Orthoses help to compensate for lower limb weakness and fatigability, thereby assisting with balance and mobility (10,36). However, orthotic devices and bracing should be considered only in patients whose foot can be held in a neutral plantigrade position (20). In a survey-based study of 314 patients with CMT, the majority of participants reported overall satisfaction with their devices. Reported drawbacks included pain and irritation after long wear, unsightly appearance, damage to clothing, and difficulty taking on and off the orthotic devices (36).

It was previously thought that exercise and strength training in patients with CMT may accelerate the rate of muscle deterioration. This idea was challenged by Burns et al (37), who conducted a 12-week intensive and progressive resistance program involving a 15-year-old patient with CMT, with the goal of strengthening dorsiflexion at the ankle. By the end of the program, the patient demonstrated improvement in isometric dorsiflexion, inversion, and walking with a decrease in foot drop. She also reported decreased pain and sensory sensation in the feet. The authors found no effect on balance or endurance (37).

### Surgical Intervention

Early surgical intervention may minimize the progression of cavovarus deformity and prevent the development of degenerative arthrosis (20). The goal of surgery is to create a more stable plantigrade foot to preserve mobility and mini-

mize bracing. To accomplish this, the surgeon must balance muscle pull by using tendon transfers, release soft-tissue contractures, and realign the weight-bearing planes of the foot with wedge osteotomies. (5,17,20,29) An explanation of the most common procedures and their purposes can be found in Table 3. Surgical arthrodesis should be a last resort and reserved for patients with advanced arthritic changes and irreducible joints (20). Figure 7 shows an example of long-standing untreated cavovarus deformity in a patient with CMT that necessitated arthrodesis.

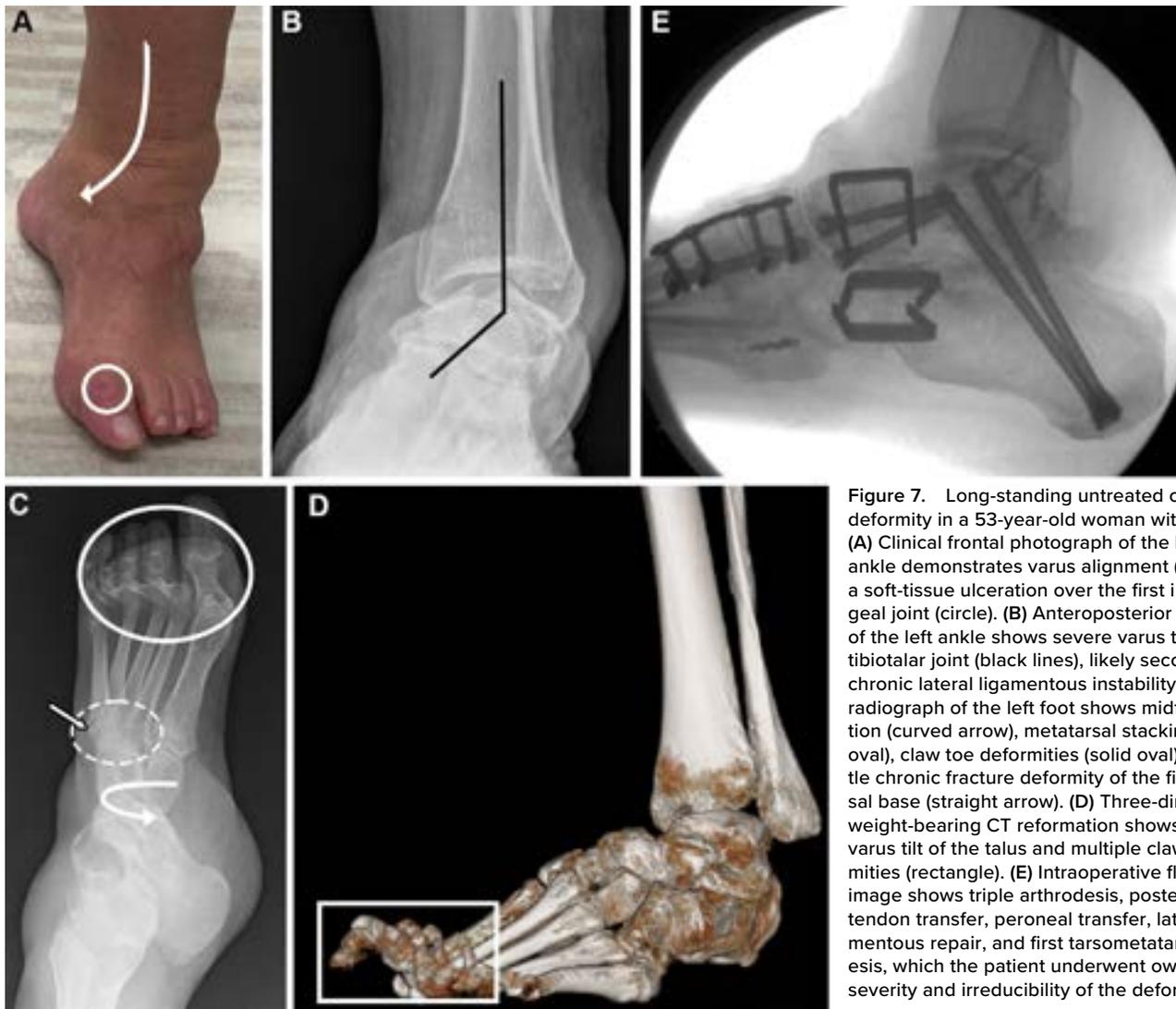
### Postoperative Imaging and Complications

The purpose of imaging in the postoperative setting is for radiographic hardware evaluation including determining hardware integrity, migration, evidence of loosening, or presence of perihardware fracture. Subtle radiographic findings include lucencies in the cuneiforms from prior posterior tibial tendon, extensor hallucis longus, or extensor digitorum longus tendon transfers. MRI in conjunction with radiography can help identify surgical complications including osteomyelitis. Examples of common postoperative imaging findings and potential complications can be found in Figures 8 and 9.

### Conclusion

CMT is the most common inherited motor and sensory neuropathy, affecting up to 0.04% of the population (1). It manifests as progressive length-dependent muscle weakness and atrophy, resulting in a characteristic complex cavovarus deformity of the foot and ankle. This deformity leads to instability and frequent falls, thus hindering patient mobility. It has been suggested to be one of the most important contributors to physical and emotional quality of life for these patients (3).

The role of the radiologist is to recognize early signs of the deformity and to identify common pathologic conditions



**Figure 7.** Long-standing untreated cavovarus deformity in a 53-year-old woman with CMT. (A) Clinical frontal photograph of the left foot and ankle demonstrates varus alignment (arrow) and a soft-tissue ulceration over the first interphalangeal joint (circle). (B) Anteroposterior radiograph of the left ankle shows severe varus through the tibiotalar joint (black lines), likely secondary to chronic lateral ligamentous instability. (C) Oblique radiograph of the left foot shows midfoot supination (curved arrow), metatarsal stacking (dashed oval), claw toe deformities (solid oval), and a subtle chronic fracture deformity of the fifth metatarsal base (straight arrow). (D) Three-dimensional weight-bearing CT reformation shows the severe varus tilt of the talus and multiple claw toe deformities (rectangle). (E) Intraoperative fluoroscopic image shows triple arthrodesis, posterior tibial tendon transfer, peroneal transfer, lateral ligamentous repair, and first tarsometatarsal arthrodesis, which the patient underwent owing to the severity and irreducibility of the deformity.



**Figure 8.** Postoperative images in three patients with CMT. (A) Anteroposterior radiograph of the foot in a 23-year-old woman shows a healed first metatarsal osteotomy with cerclage wire fixation (dashed circle), subtle ovoid lucency in the middle cuneiform (solid circle) from prior posterior tibial tendon transfer, and chronic cortical thickening of the fifth metatarsal (arrow). (B) Oblique radiograph of the foot in a 19-year-old man shows a healed first metatarsal osteotomy (dashed circle), first interphalangeal joint arthrodesis with screw fixation (rectangle), and subtle ovoid lucency in the middle cuneiform (solid circle) from prior posterior tibial tendon transfer. (C) Lateral radiograph of the foot in a 17-year-old adolescent boy shows first metatarsal osteotomy with screw and cerclage wire fixation (circle) and calcaneal osteotomy with screw fixation (arrow).



**Figure 9.** Postoperative complication in a 21-year-old man with CMT who had recently undergone surgical correction. (A) Anteroposterior radiograph of the foot shows first interphalangeal arthrodesis (rectangle), first metatarsal osteotomy (circle), and midfoot osteotomy secured with surgical staples (arrow). There was osteolysis of the fifth metatarsal head, fifth proximal phalangeal base, fifth middle and distal phalanges, fourth metatarsal head, and fourth proximal phalangeal bases, with adjacent soft-tissue swelling concerning for osteomyelitis (oval). (B) Long-axis short-T1 inversion-recovery MR image through the foot shows bone marrow edema-like signal intensity on both sides of the fourth and fifth metatarsophalangeal joints, compatible with osteomyelitis and septic joints (oval). There is a small soft-tissue abscess dorsal to the distal fifth metatarsal (black arrow) and extensive adjacent subcutaneous edema (white arrows). (C) Short-axis T1-weighted MR image through the metatarsals shows replacement of the fat signal intensity in the fourth and fifth metatarsal shafts with associated cortical destruction (rectangle), confirming the diagnosis of osteomyelitis. There is skin thickening and extensive infiltration of the dorsal subcutaneous fat compatible with cellulitis (arrows).

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associated with it, namely cortical thickening and fractures of the fifth metatarsal, ulceration and callus formation over the lateral foot, contractures of the plantar fascia and Achilles tendon, tibiotalar arthrosis, flexor tendon injury, and osteomyelitis. It is important to recognize the typical postoperative imaging appearance after surgical correction and to identify potential complications that may arise.

CMT is not the only cause of a cavovarus foot, and the information presented in this article can be used in the evaluation and treatment of other neuromuscular disorders that may result in a similar deformity.

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# QUESTIONNAIRE

## A2(25) Part 1 of 2

### Charcot-Marie-Tooth Disease of the Foot and Ankle: Imaging Features and Pathophysiology

#### INSTRUCTIONS

- Read through the article and answer the multiple-choice questions provided below.
- **Some questions may have more than one correct answer;** in which case you must mark **all** the correct answers.

#### Abstract

**Question 1:** What is the most common manifestation of Charcot-Marie-Tooth (CMT) disease?

- A:** Severe pain
- B:** Length-dependent motor and sensory deficiencies
- C:** Rapid muscle atrophy
- D:** Short stature

**Question 2:** Which deformity is widely considered the most debilitating symptom of CMT disease?

- A:** Flat feet
- B:** Hammer toes
- C:** Cavovarus deformity of the foot and ankle
- D:** Clubfoot

**Question 3:** Which imaging modality is crucial for the evaluation and treatment of the cavovarus deformity in CMT disease?

- A:** MRI
- B:** Ultrasound
- C:** Radiography and weight-bearing CT
- D:** PET scan

**Question 4:** What is a common complication associated with the cavovarus foot?

- A:** Plantar fasciitis
- B:** Ulceration over the interphalangeal joints
- C:** Osteosarcoma
- D:** Gout

#### Introduction

**Question 5:** Is it **TRUE** that the cavovarus deformity gradually leads to early-onset arthritis?

- A:** YES
- B:** NO

**Question 6:** Patients with CMT usually present with a pes cavus deformity that is .....

- A:** Forefoot driven
- B:** Idiopathic
- C:** Hindfoot driven

#### Clinical Presentation

**Question 7:** Which muscles are most commonly affected first in CMT disease?

- A:** Intrinsic foot muscles
- B:** Calf muscles
- C:** Gluteal muscles
- D:** Hand muscles

**Question 8:** Which of the following muscles are spared?

- A:** Peroneus brevis
- B:** Posterior tibialis
- C:** Gastrocnemius
- D:** Soleus

**Question 9:** Is it **TRUE** that a majority of patients with CMT will demonstrate similar severity of muscle involvement in both limbs, but up to 5% may display significant asymmetry, a finding associated with more severe disease?

- A:** YES
- B:** NO

**Question 10:** The most common presenting concerns are .....

- A:** Frequent falls
- B:** Foot pain and cramping
- C:** Ankle sprains
- D:** Altered gait

**Question 11:** What is the reported prevalence of CMT disease in patients who present with bilateral cavovarus feet?

- A: 25%
- B: 50%
- C: 79%
- D: 91%

**THE END**

# QUESTIONNAIRE

## A2(25) Part 2 of 2

### Charcot-Marie-Tooth Disease of the Foot and Ankle: Imaging Features and Pathophysiology

#### INSTRUCTIONS

- Read through the article and answer the multiple-choice questions provided below.
- **Some questions may have more than one correct answer;** in which case you must mark **all** the correct answers.

#### Imaging Features

**Question 1:** Which of the following is a key imaging feature to recognise in CMT-related cavovarus deformity?

- A:** Increased bone density
- B:** Shortened calcaneus
- C:** Abnormal Meary angle
- D:** Widened joint spaces

**Question 2:** In what image of figure 2, is the sinus tarsi (see-through) sign and the double talar dome sign seen?

- A:** A
- B:** B
- C:** C
- D:** D
- E:** E

**Question 3:** What is the role of weight-bearing CT in evaluating patients with CMT disease?

- A:** To assess bone density
- B:** To evaluate hindfoot alignment
- C:** To identify tumours

**Question 4:** Which imaging modality is primarily used to assess soft-tissue pathologic conditions associated with cavovarus deformity in CMT disease?

- A:** CT scan
- B:** Radiography
- C:** MRI
- D:** PET scan

**Question 5:** Is it **TRUE** that the authors of a study discovered that patients with CMT often demonstrated paradoxically normal calcaneal pitch angles secondary to severe midfoot supination, which overwhelmed the dorsiflexion of the calcaneus?

- A:** YES
- B:** NO

#### Treatment

**Question 6:** What is the recommended first-line treatment for patients with mild CMT deformity?

- A:** Surgical intervention
- B:** Ground-reaction ankle foot orthoses
- C:** Physical therapy only
- D:** Custom shoes

**Question 7:** Is it **TRUE** or **FALSE** that exercise and strength training in patients with CMT may accelerate the rate of muscle deterioration?

- A:** TRUE
- B:** FALSE

**Question 8:** What is the main goal of surgery in patients with CMT disease?

- A:** To reduce muscle atrophy
- B:** To enhance physical appearance
- C:** To create a more stable plantigrade foot

**Question 9:** What is one of the surgical procedures used to correct the deformity in CMT disease?

- A:** Joint replacement
- B:** Wedge osteotomy
- C:** Ligament reconstruction
- D:** Nerve grafting

**Question 10:** What surgical procedure is considered a last resort in the treatment of cavovarus deformity in CMT disease?

- A:** Tendon transfer
- B:** Soft-tissue release
- C:** Osteotomy
- D:** Arthrodesis

**THE END**



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**ANSWER SHEET**

**A2(25)**

Charcot-Marie-Tooth Disease of the Foot and Ankle Imaging Features and Pathophysiology

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