Clinics in Surgery

Case Report
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A Rare & Complex Case of Neuro-Pathic Diabetic Foot-Charcot Marie Tooth Disease

Khalid MS, Akram R, Goel R, Elmetwally A, Elsantawy H, Khan Waheed A, Abdel Haliem A, Ahmed AM and Buxton J*

Department of Vascular Surgery, Royal Blackburn Hospital, Blackburn, UK

Abstract

Charcot Marie Tooth (CMT) disease is an inherited peripheral nerve disorder that causes abnormalities in the nerves that supply feet, legs, hands and arms with both motor and sensory systems component. The clinical presentation includes numbness, tingling, burning sensation, muscle wasting and loss of temperature in the peripheries and feet ulcers that sometimes mistaken as peripheral vascular disease. If CMT has associated with diabetes myelitis, it presents as a severe form of the disease.

Keywords: Charcot-Marie-tooth; Peripheral; Neuropathy; Hereditary; Neuropathic ulcers

Case History

We are going to present a 57-year-old male who presented with a complex symptom of pain, redness, paranesthesia, poikilothermia and multiple small trophic ulcers on both feet. This patient had a family history of Charcot-Marie-Tooth disease besides he was suffering from type 2 DM. This patient started symptoms of this disease in his adolescence. He was referred to vascular clinic to evaluate peripheral vascular disease of his both legs and feet. The combination of the Clinical examination along with non-invasive tests like ABPI and Duplex scan confirmed this patient has got no peripheral vascular disease rather neuropathic condition secondary to CMT disease (Figures 1-5) [1-8].

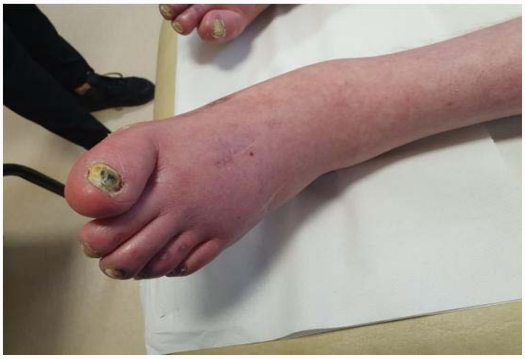

Figure 1: Left foot of the patient.

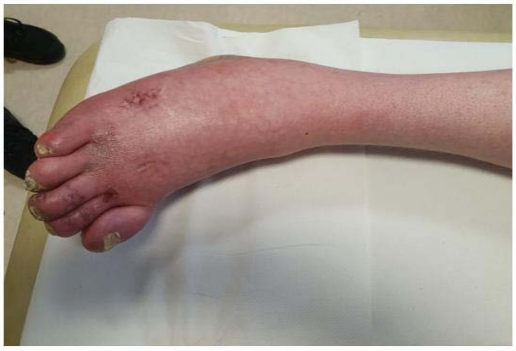

Figure 2: Right foot.

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Correspondence:
Sufian Khalid M, Department of Vascular Surgery, Royal Blackburn Hospital, Blackburn, UK, E-mail: sufian999@hotmail.com

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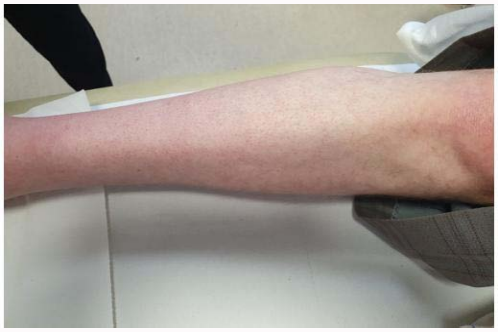

Figure 3: Right leg.

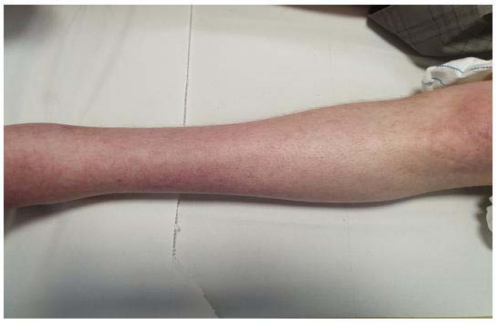

Figure 4: Left Leg.

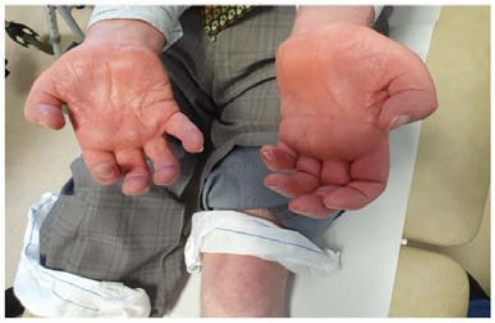

Figure 5: Hands.

Discussion

CMT is a complex nerve condition that was named after three doctors who initially mentioned this disease in 1886. 1 Jean Martin Charcot, 2. Pierre Marie, 3. Howard Henry Tooth of the UK. The term CMT is considered as being similar to hereditary motor sensory neuropathy condition also known as HMSN.

Charcot Marie Tooth infection is brought about by inherited genetic mutations engaged with the function and the structure of the peripheral nerves. The general predominance of CMT is roughly 19 occasions for each 100,000 individuals which can differ in between. Depending upon the kind of CMT, start can be from birth to adulthood and development is typically slow. CMT is not usually a life-threatening condition, and it very rarely involves the brain.

The clinical features of CMT: The important manifestations of CMT either regularly comes up in younger age or early adulthood which may consist of weakness and possible wasting of muscles for the foot, lower leg, hand, and lower arm. Patients may have a decreased sensation in the fingers, toes, arms and legs.

Other clinical findings include the abnormalities in the muscles of the calf and foot which can present as high arches and hammertoes. A patient may experience issues using the fingers and the hands. They may suffer from unsteady ankles and issues with balance. Many experience cramps in the forearms and the lower legs. While some have vision and hearing loss. Some experience posture abnormalities like scoliosis. Reflexes may be decreased. Nature of the symptoms and their seriousness may vary essentially among the people, even among siblings or other relatives who have inherited this disease.

In the early phases of life, many probably won't realize that they have CMT, on the grounds that the complications are insignificant or very mild. The patient who may have CMT, need to see a neuro-physician and a geneticist for additional examinations.

Diagnosis of CMT

1. Nerve conduction examination for peripheral neuropathy: This measures the speed and the strength of the electrical signs that penetrate the peripheral nerves. The electrodes are applied to the skin which distributes small electric currents that stimulate nerves. A week or delayed response may suggest an abnormal nerve condition or a high probability of CMT.

2. A biopsy: Usually a little piece of peripheral nerve normally from the calf is taken and is assessed and tested in a lab which can confirm the presence of CMT.

3. Electromyography (EMG): This involves putting a tiny needle into the diseased muscle. As the patient contracts or relaxes the muscle, an electric activity is measured. Evaluating various muscles will indicate which ones are experiencing the condition.

4. Hereditary/Genetic testing: A blood test is taken to evaluate whether the individual possesses the diseased genes or not [9,10].

D/D of Neuropathic and Ischemic Ulcers

It is very important to understand the difference between neuroischemic (Ischemic) and neuropathic ulceration, to differentiate the vascular elements, we perform the following tests and investigations which are mainly non-invasive i.e. ABPI, TBPI, TcPO2, Doppler ultrasound waves and duplex US scan. Neuropathic ulcers occur because of peripheral neuropathy, regularly in diabetic cases where Local paresthesias or the lack of sensation, over the pressure points of the foot can lead to extensive microtrauma, breakdown of overlying tissue, and inevitable ulceration. Likewise, neuropathy can bring about minor cuts or scrapes unsuccessfully treated or managed which can eventually turn into ulcers. Peripheral sensory neuropathy brings about loss of protective sensation that enhances the risk of repetitive injury or acute trauma leading to ulceration. Motor neuropathy influences the muscles needed for foot development and normal movement. The abnormal muscles and bony structure distribute the forces abnormally during walking which can cause reactive thickening of the skin, or callus formation, at the pressure points of the foot. Ischemic tissue necrosis underneath the callus turns into the breakdown of skin and subcutaneous tissue, leading to a neuropathic ulcer with an appearance that is punched out (Tables 1-3) (Figure 6).

Management of the Neuropathic Ulcer

The following safety measures can eliminate or decrease the

Table 1: Symptoms and signs of neuropathic versus ischemic ulcers.

Neuropathic Ulcer	Ischemic Ulcer
Painless	Painful
Normal pulses	Absent pulses
Regular margins	Typically punched-out appearance, irregular margins
Often located on plantar surface of foot	Commonly located on toes
Presence of calluses	Calluses absent or infrequent
Loss of sensation, reflexes and vibration	Variable sensory findings
Increase in blood flow (AV shunting)	Decrease in blood flow
Dilated veins	Collapsed veins
Dry, warm foot	Cold foot
Bony deformities	Possibly no bony deformities
Red appearance	Pale, cyanotic

Table 2: Typical features of ulceration.

	Neuropathic	Ischemic
Pulses	Bounding	Diminished/absent
Pain	None/minimal	High degree of pain
Location of Ulceration	Pressure areas	Borders of feet
Callus	Often large amounts	Minimal

Table 3: Neuropathic foot & Neuroischemic foot.

Characteristics	Neuropathic foot	Neuroischemic foot
Skin temperature	Warm	Cold
Pain	Painless	Painful
Skin Colour	Not altered	Dependent rubor
Callus	Thick at pressure point	Usually not present
Ulcer	Usually on tips of toes & plantar surfaces under metatarsal heads	Often on margin of foot, tips of toes, heels
Peripheral pulses	Bounding	Feeble/absent
ABI	More than 0.9	Less than 0.9
Complication	Charcot Joints	Critical ischemia

risk of creating the neuropathic ulcers in a high-risk patient as well as it reduces the complication of someone who is already having neuropathic symptoms. Consider normal podiatric care to eliminate excessive calluses and monitor for likely ulcerations. Perform regular check-ups of the feet for any abnormal changes in the Colour or experiencing pain or callous formation. Ensure the footwear is properly fitted which will also help to avoid the pressure point from rubbing. Protect both feet from any injury, accident, infection and extreme temperatures. Never walk shoeless. Always wear slippers or shoes while in the house. Stay away from splashing feet. Insensate feet can be scalded easily without the patient's knowledge. Manage diabetes or other applicable health conditions to expedite the healing process one of the most fundamental components to promote effective healing of the neuropathic ulcers is to decrease pressure on the affected and diseased areas. Notwithstanding, alleviating pressure from the injury should be balanced with providing appropriate circulation to the extremities, so too much bed rest isn't advised. Contact casts can be utilized to reduce the pressure of the affected region while permitting the patient to stay ambulatory. Therapeutic shoes can be provided to serve the same function, but they are usually used to prevent or to avoid recurrence.

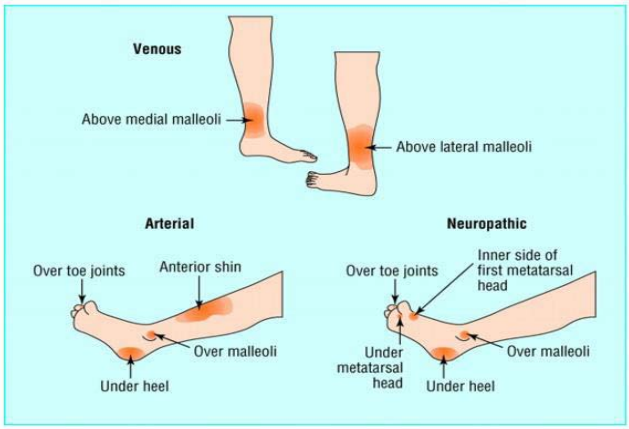

Figure 6: Leg ulcers.

If the ulcer doesn't heal after all the conservative measures, then surgery may be considered to correct the deformities in the foot which help to remove excessive pressure.

The wound should thoroughly clean and debrided down to the healthy, normal and bleeding tissue. Usually there is an infection beneath the superficial necrotic tissues which can involve the bone and the bone marrow. In these cases, debridement of the dead bones is important for successful healing. It is recommended for the wound environment while healing to be moist but also having good oxygenation. The specific properties of the dressing ought to be matched with those of the injury.

Conclusion

Charcot-Marie-Tooth disease is a hereditary disorder which is characterized by sensory & motor polyneuropathy with foot deformity, sensorineural hearing loss, developmental delay and walking problems. The diagnosis of this condition depends upon physical symptoms, medical & family history and neurological examination & tests. It can present muscle weakness in the arms, hands, legs and feet. There is associated decreased muscle bulk, sensory loss and decreased tendon reflexes. Besides this, patients can present with neuropathic ulcers commonly on the feet & occasionally other parts of the body. It is very important to recognize this condition early, evaluate it properly and treat with multidisciplinary approach which may help patient to have a better quality of life.

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