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Ankle Fracture in Hereditary Sensory Neuropathy Type 1

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ABSTRACT

Ankle fractures account for approximately 9% of all adult fractures annually. The ankle anatomically is particularly vulnerable to significant skin compromise in the setting of trauma. Significant fracture blistering and soft tissue compromise is predominantly seen in high-energy ankle injuries. Hereditary sensory autonomic neuropathy type I is a rare progressive neurological disorder resulting in distal sensory loss and autonomic disturbances with variable motor involvement. We present a case involving a hereditary sensory autonomic neuropathy type I patient with unexpected significant soft tissue injury on the background of a low energy ankle fracture. The aim was to outline the diagnosis and complex management considerations related to hereditary sensory neuropathic-associated ankle injuries.

Case Report

At the beginning of March 2019, a 47-year-old patient with a background history of HSN I fell from the step of a pickup truck, approximately 2 m off the ground, sustaining a closed simple Weber B ankle fracture (Figs. 1 and 2). He was assessed and splinted in a community hospital by the emergency department roughly 6 hours after sustaining the injury. It was noted in the community hospital documentation that the patient had some evidence of a superficial abrasion on the medial malleolus with no mention of fracture blisters or widespread skin or soft tissue compromise.

The patient was then transferred via ambulance to the main tertiary care center for further orthopedic care. Due to hospital-to-hospital transfer protocols, there was a further delay of 4 hours to being seen by the orthopedic team. Upon a ten-hour delay to tertiary care, the patient had developed a widespread soft tissue injury similarly seen in high-energy plafond injuries (Fig. 3). On clinical exam, this patient had altered sensation globally in his distal extremity. Additionally, he had bilateral longstanding foot drop, a finding common in patients with HSN I.

He was taken to the operating room, within 24 hours of admission, for treatment of his ankle injury with a temporizing ankle spanning external fixator (Figs. 4 and 5). The application of the external fixator allowed the team to monitor his soft tissues in anticipation for definitive open reduction internal fixation once the soft tissue compromise had subsided. The plastic surgery service was consulted to provide assistance in managing his significant soft tissue injury (Fig. 6). Hemorrhagic blistering developed rapidly during the first days of his admission to the tertiary care hospital. The hemorrhagic blisters were...
de-roofed and silver sulfadiazine dressings were applied under the direction of the plastic surgery team.

The wound care team then transitioned to jelonette dressing and collagenase ointment, which was applied daily to treat the soft tissue injuries. The patient developed necrotic tissue to the medial malleolus, which required hydrofera blue dressings. Inadine dry dressings were also applied to settle the soft tissue envelope.

At the end of April 2019, the patient unfortunately developed a pin site infection in the lateral calcaneus pin. The external fixator was removed and irrigation and debridement were performed after 2 months since his initial operation. He was then treated in a cast with a vacuum-assisted dressing over his lateral calcaneal pin site wound, as his overall soft tissues were at high risk for further breakdown if open reduction internal fixation were performed at that time.

The Infectious Disease team was consulted in the interim to tailor his antibiotics to treat his infected pin site. Due to his significant soft tissue compromise on the background of the underlying hereditary sensory neuropathy, it took over 4 months for the soft tissue compromise to fully resolve before being able to safely perform open reduction internal fixation of his ankle injury. The patient received definitive ankle fixation in June 2019. The operative team utilized locking plate technology with syndesmotic screw fixation (Figs. 7 and 8). Expectedly, this proved to be a challenging operative procedure given the length of time since his fracture and his vulnerable soft tissue bed. The patient was followed up at 2 weeks and 6 weeks post definitive surgery. His soft tissues, after months of significant wound care management from the multidisciplinary team, demonstrated significant improvement at
clinic follow up (Figs. 9 and 10). The patient was transitioned to full weightbearing as tolerated at his 9-month follow-up in November 2019. With the ongoing COVID-19 pandemic, a telephone follow-up was conducted at 16 months postfracture in June 2020. The patient reported a full recovery with complete healing of his soft tissue injury.

Discussion

This case highlights the importance of a multidisciplinary team to manage the challenges in patients with HSN I. This patient required orthopedic surgery, plastic surgery, the infectious disease team and the local wound care team to address the complexity of this "simple" ankle fracture. A thorough history, clinical examination, and early involvement of the multidisciplinary team are paramount in patients who present with orthopedic trauma on the background of neuropathy. The significant duration of his treatment for what initially appeared as a routine ankle fracture is a testament to the complexity of his underlying neuropathy.

Dyck classified hereditary sensory neuropathies in 1993. There are 5 types (HSN I-V). HSN I is the most common subtype, inherited in an autosomal dominant pattern. HSN I has been linked to SPTLC1 and RAB7 genes, usually presenting with familial heritance patterns. HSN I is characterized by late onset presentation with progressive neurological findings. Diagnosis is based on clinical signs, symptoms and family history. Progressive sensory, vasomotor function, and motor loss in the distal extremities are classic findings of this condition. Sensory potentials are usually completely absent in the lower extremity in comparison to the upper extremity which can be found to be normal. In nerve studies, axonal nerve damage leads to slowing of motor conduction and demyelination. Sural nerve biopsies conducted in patients with the SPTLC1 mutation have demonstrated very limited myelinated fibres remaining.

With neuropathic ulcer formation and sensory dysfunction in the feet common, HSN I has been nicknamed "pseudodiabetic foot syndrome." Timely operative management, consideration of strict extended non-weightbearing orders postoperatively and attentive soft tissue management in this population is crucial. This has been consistently recommended in the literature surrounding the treatment of the diabetic ankle fracture population. A study in the diabetic ankle fracture population with early protected weightbearing at 2 weeks post-open reduction internal fixation recorded a complication rate of 25%. The insensate foot, much like other neuropathies, such as diabetic neuropathy, predisposes this cohort to high risk of skin breakdown, mal-union, failure of hardware and possible amputation. Delaying ankle fracture surgery had a 12.9% of wound complications versus 3.6% in the expedited group in a systematic review. Nonoperative management in the diabetic literature has been associated with a staggering 75% complication rate. Patients with HSN I require early surgical management to avoid devastating complications seen in similar neuropathies.

This patient presented with a severe soft tissue injury on the background of a low energy ankle fracture. Around 6.6% of ankle fractures...
present with fracture blisters, on average delaying definitive operative management for 6 days. The lower leg is a vulnerable location for soft tissue injury given its unique anatomical characteristics of limited subcutaneous tissue, arborizing venous system, and flat dermal papillae. Blisters can develop as early as within 6 hours of the fracture, making timely orthopedic intervention integral to managing the evolving soft tissue injury. The fracture pattern does not always correlate with the severity of the soft tissue fracture blistering that can develop, as demonstrated in this case.

The disruption of the epidermis and dermis by energy dissipated during a trauma leads to the development of fracture bullae. Tissue hypoxia along with a combination of torsional and shear forces are the underlying mechanisms for fracture blister formation. There are 2 classified types of fracture blisters. Clear fluid filled blisters represent injury to the dermis layer with some remnant epidermal cells, which allow a quicker time to epithelization. Blood filled blisters represent complete disruption of the dermal-epidermal junction. Complete resolution of this type of blistering has been reported to take up to 16 days post injury.

Patients with HSN I have an abnormal underlying soft tissue envelope. Skin biopsies of patients with HSN I demonstrate lack of innervation to the sweat glands. Due the autonomic dysfunction, patients with HSN I can experience anhidrosis, hypoanhidrosis, or hyperhidrosis. The lack of normal tissue response to trauma in patients with HSN I likely contributed to the severity of this patient’s soft tissue envelope injury, the delayed resolution of his numerous blood-filled fracture blisters, and the subsequent skin necrosis that he experienced.
Involvement of the plastic surgery or wound care team is vitally important. Strauss et al presented de-roofing blisters and twice daily application of silver sulfadiazine until re-epithelization as a successful treatment protocol for fracture blisters in lower limb orthopedic trauma. The advent of negative pressure wound therapy has also been a welcomed addition to the armament necessary to treat extensive fracture blistering. Negative pressure wound therapy dressings have been demonstrated to improve tissue perfusion, reduce soft tissue edema and stimulate epithelization of wound beds. This case demonstrated effective use of both of these techniques.

In conclusion, this patient presented with a bread and butter orthopaedic problem that resulted in an extensive treatment course. The case presented highlights the fact that not all ankle fractures behave the same. Surgeons should proceed with caution when treating simple ankle fractures, as they can be deceiving in patients with neuropathic conditions like HSN I. Orthopaedic trauma in hereditary neuropathy patients represents a challenge requiring early intervention with a multidisciplinary approach. Failure to recognise this could lead to misdiagnosis, mismanagement and adverse outcomes.

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