

A rare case of neurofibromatosis type I

Mary Eagle is Community Tissue Viability Adviser, Blackwater Valley and Hart Primary Care Trust and Shiu-Ling Briggs is Tissue Viability Adviser, Royal Surrey County Hospital, Guildford and Waverley Primary Care Trust

Neurofibromatosis type I (NFI), sometimes referred to as Von Recklinghausen neurofibromatosis, is an inherited disorder characterised by the formation of tumours (neurofibromas) involving nerve tissues in the skin, subcutaneous tissue, cranial nerves, and spinal root nerves (Riccardi and Eichner, 1986). NFI is inherited as an autosomal dominant trait, affecting one in every 2,500 people worldwide including 25,000 in this country alone. The disease occurs in approximately one in every 3,000 children. NFI sometimes also appears in families with no previous history of the condition, as a result of a new genetic mutation. In NFI the abnormality lies in one of the genes encoding a protein called neurofibromin. This results in unchecked growth of neurofibromatous tissue in nerves, which can put pressure on them causing pain and severe nerve damage (Riccardi and Eichner, 1986). It also leads to loss of function in the area served by the nerve. Problems with sensation or movement can occur as a result, depending on the particular nerves affected. With multiple neurofibromas, the tumours may result in bumps under the skin; the subcutaneous lumps are characteristic of the disease and increase in number with age. Although usually benign, neurofibromas can sometimes degenerate into cancer.

Symptoms

Symptoms can be extremely varied, depending on the age of the patient and severity of the disease and may include:

- ▶▶ Rubbery soft nodules/tumours under the surface of the skin called nodular neurofibromas
- ▶▶ Large, soft, infiltrating tumours that spread out under the skin. These are called plexiform neurofibromas, which may be darker in colour than the surrounding skin and can cause disfigurement
- ▶▶ Multiple soft tumours visible on the skin or deeper in the body, which can be viewed

by scanning. Magnetic resonance images (MRI) are useful to evaluate mediastinal masses, spinal cord tumours, deep plexiform neurofibromas, abdominal and pelvic lesions and neurofibromas of the brachial or sacral plexus

- ▶▶ Pain (associated with affected peripheral nerves)
- ▶▶ Attention deficit hyperactivity disorder in a minority of patients (Schrimsher et al, 2003) along with mild impairment of intellectual function, however; 60% of those with the condition have specific learning difficulties and behavioural problems. Very few children with neurofibromatosis have a below normal IQ and most are outwardly bright and lively but, at school, they have particular trouble with reading, writing and maths. They may lack coordination, have poor spatial judgment, problems with short-term memory and act on impulse
- ▶▶ Fracture of the long bones of the leg in early childhood. Skeletal abnormalities such as scoliosis or bowing of the legs might occur, resulting in fractures. Congenital pseudoarthrosis may be apparent at birth. Bowing of the tibia is the commonest presentation. Thinning and angulation of long bones with prominence of the anterior tibia and progressive deformity can occur throughout early childhood. Bowing of the forearm is less common
- ▶▶ Café au lait spots (pigmented birthmarks). Six or more of these form one of the diagnostic criteria, but are not essential for diagnosis
- ▶▶ Freckles in the underarm, underbreast or groin
- ▶▶ Convulsions, the frequency of which vary according to the extent of the disease
- ▶▶ Lisch nodules (haematomas of iris) and/or tumours on the optic nerve, also known as an optic glioma, can cause visual impairment and /or blindness.

Treatment for tumours

There is no specific treatment for neurofibromatosis (Arun and Gutmann, 2004). Tumours that cause pain or loss of function are removed on an individual basis. Tumours

Table 1

Medication at time of admission

Drug	Dosage
Amitriptyline	75mg once daily
Diazepam	2mg three times a day
Temazepam	4mg once a night
Co-dydramol	2 tablets every 4–6 hours as required
Warfarin	2mg/1mg, titrated doses according to blood profile of clotting time
Aspirin	75mg once daily

that have grown rapidly are usually removed promptly as they may become malignant.

Case report

The patient was a 62-year-old woman with mild learning difficulties and obesity, who was married and lived with her husband who was also her main carer. The patient had been diagnosed with multiple sclerosis and cervical demyelination 19 years previously. She also suffered from recurrent episodes of pulmonary embolism (PE) and so had had an intravenous cable filter inserted a year earlier to reduce the risk of further emboli. The patient also had rheumatoid arthritis and severe back pain which resulted in reduced mobility.

Medical history showed that the patient had suffered from extensive NFI for many years. A neurofibroma lesion on her left thigh has been slowly enlarging for 9 years. This caused increased pain and further reduced her mobility. She had been referred to a plastic surgeon 9 years previously to have the tumour on her left thigh debulked. However, she and her husband, having considered the effect of the surgical option, declined because they could see no long-term benefits.

The patient remained under the close
Wounds UK, 2008, Vol 4, No 3

supervision of her medical consultant at the hospital because of her NFI diagnosis, her poly-pathology and interrelated diseases. care of which was shared between her local primary and secondary care units.

The patient presented at A&E following two days of lower chest and pleuritic pain that worsened and became sharp on inhalation. A provisional diagnosis of suspected recurrent pulmonary embolism due to the failure of anticoagulant therapy to reduce her risk of developing intravenous clots was made. The patient's medication regimen at the time of admission is listed in *Table 1*.

Weeks 1 and 2 of hospital admission

The patient was admitted to the medical ward for the treatment of a recurrent pulmonary embolus, monitoring of blood clotting profile and observations.

On the third day after admission, the ward nurses noticed a haematoma developing in a neurofibromous tumour on the posterior aspect of the left thigh. The patient was found sitting by the side of the bed, unaware that she was bleeding, with a pool of blood under the chair. The bleeding from the haematoma looked fresh with active oozing from several small areas. It was estimated that 500ml of blood had been lost. A low adherent dressing and pad was applied immediately, and secured with a bandage applying gentle pressure to the bleeding wound. Care was taken not to apply too much pressure with the bandage due to the potential tourniquet effect around the thigh. Warfarin (anticoagulant for prevention of the patient's thromboembolic conditions) and enoxaparin (used in the treatment of pulmonary embolism) were stopped due to their anticoagulant properties. An intravenous cannula was sited in order to ensure immediate access for fluid and blood replacement. Observations of temperature, pulse, respiration and blood pressure were recorded for the next 24 hours.

The patient's blood pressure reduced from 160/100 to 100/60 within 24 hours and the bleeding continued. A unit of blood was transfused and blood tests for sepsis screening



Figure 1. The wound at presentation. Three days post-operation, the wound was necrotic and sloughy, showing signs of continued infection.

were sent for microbiological studies.

Forty-eight hours later the bleeding stopped and the patient was haemodynamically stable. At this stage enoxaparin (40mg twice-daily) was recommenced. The next day the patient spiked a temperature of 39°C and felt generally unwell. Infection of the haematoma was diagnosed, and the patient was advised that evacuation of the infected haematoma under general anaesthetic was necessary. However, she initially refused to accept the necessity for surgical treatment and did not appreciate the severity of the problem. This could possibly have been due to her mild learning disability.

The patient was incontinent of urine and faeces as a result of the difficulty she had getting onto a bed pan due to pain, immobility and her obesity. She also refused consent for urinary catheterisation, however, after careful discussion, she consented as the haematoma site was becoming contaminated with urine. Flucloxacillin was also given intravenously and a further unit of blood was transfused as her haemoglobin level had dropped to 5.3g/dl.

The following day the patient gave her consent for the evacuation of the haematoma. The incisional wound was extensive, measuring 22cm x 20cm. The area was 'laid open' as there was insufficient skin to obtain full closure and the wound was left to heal by secondary intention. Following the instructions of the consultant, a low-adherent dressing was applied to the wound by the ward staff with a secondary dressing to control any strike-through of exudate. Post-operatively her temperature remained elevated and she remained catheterised, but continued to be incontinent of faeces, which was further contaminating the wound. Systemic antibiotics (ciprofloxacin and trimethoprim) continued to be given to control infection of the haematoma site.

The patient was referred to the tissue viability adviser (TVA) at the acute hospital on the third post-operative day. The wound was extensive, necrotic and sloughy, showing signs of continuing infection of the haematoma site (*Figure 1*). There was an area of erythema around the wound that was 5cm wide. Intravenous antibiotics were continued as microbiology reported that coliform and methicillin-resistant *Staphylococcus aureus* were cultured. It was agreed among the members of

the multidisciplinary team that further surgical debridement was required as the patient continued to spike a temperature despite taking antibiotics and because the wound bed had become covered with necrotic and sloughy tissue within 48 hours of the evacuation of the haematoma.

A plastic surgeon was consulted and he advised surgical debridement of the haematoma site followed by a skin graft, but the patient declined further general anaesthesia or surgery. Under the guidance and advice of the TVA a joint decision was taken to manage the surgical site with a primary antimicrobial ionic silver dressing (Silvercel, Johnson & Johnson Wound Management, Ascot) as a primary dressing, to reduce any bacterial bioburden (the microbial loading of the skin and/or wounds with normal commensals and potential pathogens) (White et al, 2001; White and Cooper, 2003) (Figure 2). A non-adherent silicone foam dressing (Mepilex, Mölnlycke, Dunstable) was used as a secondary dressing in order to reduce procedural pain associated with dressing changes (Meaume et al, 2003; Thomas, 2003). With this regimen the wound continued to show improvement and there was no further development of necrosis and sloughy tissue. Three weeks after surgical evacuation of the haematoma the patient was recovering sufficiently to be considered for transfer to a community hospital for continuing care and rehabilitation.

Week 3: transfer to community rehabilitation unit

In July 2005 terrorist bombings in London necessitated the clearing of suburban acute unit beds to take the overload of patients from London. As a result the patient was transferred to a primary care community rehabilitation unit two weeks after surgery (Figure 3). The patient was not mobile, still on bed rest with a large wound and was unfit for discharge home. A meeting between TVAs from primary and secondary care was arranged to ensure continuity of wound care. The patient's medical conditions were complex and medication to control her recurrent episodes of pulmonary embolism was still

being titrated. Due to these complexities her care was shared between the rehabilitation teams in primary care and consultants in the secondary care team.

The staff at the rehabilitation unit did not have the knowledge or skills to care for someone with such a large wound. The secondary care unit's tissue viability nurse (TVN) was based in the hospital setting and the primary care TVN's office was based in a community health centre 24 miles away from the rehabilitation unit, therefore daily visits were not feasible in the long term.

Ongoing wound management

The tissue on the wound bled easily if touched or disturbed. Due to its position and the wound bed's fragility, the patient was unable to be transferred using a hoist and sling; the wound was exactly where the sling needed to be positioned (Figure 3) and for a further five weeks she was confined to bed rest. She was at high risk for pressure and shear damage because of her immobility and her inability to reposition herself. Pressure area management was a major concern, as cost to the patient's quality of life was further compounded by the financial cost to the NHS with the cost of treating a pressure ulcer being approximately £400,000 per patient (Mahoney, 1999). The TVNs further collaborated with a manual handling adviser, physiotherapist, occupational therapist and nursing staff. A decision was made to nurse the patient in a four-section profiling bed that included a knee break. The knee break is designed to fit along the legs with a break at the knee to support the upper thigh and lower leg. This break in the base of the bed can be positioned to suit each individual, helping to stop the patient slipping down the bed thus preventing shear and friction damage to the skin. An airflow mattress was in situ to help prevent slipping and shear damage. Pressure ulcer prevention and management, risk assessment and reassessment was monitored and the patient was initially repositioned every two hours with the help of a glide sheet.

During the five-week bed rest the patient was re-admitted to the acute unit for three days



Figure 2. The wound after one-week of treatment with Silvercel as the primary dressing.



Figure 3. Following two weeks of dressing with Silvercel, the patient was transferred to a primary care unit.



Figure 4. The wound following five weeks of Silvercel treatment.

with a further PE. Communication between the two TVNs ensured wound management remained unchanged during her readmission and the acute unit's TVA undertook management of care during this readmission.

IV antibiotics were discontinued after her initial admission to the primary care rehabilitation unit and Silvercel was used for the duration of the wound management (Figures 4–8). Due to the high exudate levels the dressing was changed daily for the first five weeks then every third day until healing was achieved.

It is usual local clinical practice to 'clean' a wound of bacteria with a silver-impregnated dressing then move on to a non-antimicrobial dressing to progress through healing. However, this wound had the potential to remain contaminated or critically colonised until it had completely healed as the patient continued to have faecal leakage at times and still had occasional incontinence of urine after her catheter had been removed resulting in her referral to a continence adviser.

The community TVN visually reviewed and inspected the wound, assessing evaluating and reconsidering wound management daily for the first week. Visits were then reduced to weekly. As TVA visits reduced, telephone calls to the ward staff were made initially daily and then gradually reduced as the ward staff became more confident to provide care. Designated ward staff were educated and trained by the community TVA to undertake the complex dressing changes, and to observe the peri-wound skin integrity.

Involvement in care

The TVAs identified that the patient's wound would need to heal before she would be able to progress to full mobility and self care. The rehabilitation unit was designed for short stay usage to rehabilitate patients back to self-care abilities within their own limitations.

Three meetings with representatives of the multidisciplinary teams present were led by the TVAs to agree working relationships and accountabilities within the multidisciplinary teams involved in the patient's care. A multidisciplinary care package was designed and agreed. Care was focused initially around the patient's inability to mobilise due to:

- ▶▶ The size and position of the wound
- ▶▶ Potential problems involved with her previous history of multiple PE
- ▶▶ Her obesity and attitude to her general condition.

The patient and her main carer (her husband) were kept informed and agreed with all decisions made during the meetings. Medical staff involved in her care included the surgical consultant, surgical team, medical teams,

primary and secondary care tissue viability advisers, secondary and primary care nursing teams, microbiologist, haematologist, infection control team, physiotherapist, dietician, occupational therapist, equipment manager; manual handling adviser; continence adviser; medicine management team (primary and secondary care) and pastoral ministerial services.

Nutrition

Nutritional support is fundamental to patient care, and needs will vary from patient to patient. Successful wound healing and other treatment may depend on a patient's nutritional status (Williams and Leaper, 2000). The concept of malnutrition is often related to inadequate consumption of food leading to weight loss. Neno and Neno (2006) debate, however, that malnutrition can also refer to over-nutrition (the intake of nutrients in excess of requirements). There is less research into obesity and over-nutrition than under-nutrition, despite being central to public health initiatives (Department of Health, 2004a).

Some wounds may become 'stuck' at some stage during the wound healing cascade if they are regularly deprived of one or more of the essential nutrients known to fulfill roles as structural components, enzyme co-factors, or physiological mediators in skin repair and regeneration (Lansdown, 2004). It was important that this patient's wound did not fall into this chronic state. The wound's progress to healing was pivotal to her rehabilitation programme. The TVA discussed with the patient, her carer and nursing staff the importance of a 'good' well-balanced diet. Her carer often brought her calorie and carbohydrate-laden gifts, which she ate with relish. The patient was referred to a dietician who aimed to ensure a balanced diet that would aid wound healing and address her need to lose weight.

Patient attitude

Russell (2001) states the nurse is in the ideal position to make assessment of a patient's wound healing and then decide on appropriate nutrition. However, the reduction in quantity of the patient's favourite snacks made her



Figure 5. Wound bed showing hypergranulation.



Figure 6. Wound position back left thigh.



Figure 7. The wound at week 8 of treatment.



Figure 8. The wound healed after 11 weeks of Silvercel treatment.

unhappy and bored. There was a need to empower the patient to be able to realise and maintain her potential in relation to health and well-being (Department of Health, 2004b).

A person who has a degree of learning disability probably has significant reduction in the ability to understand new or complex information and a reduced ability to cope independently (Department of Health, 2001). This inability impacted on the communication and interaction between healthcare providers and the patient.

As the patient got a great deal of comfort through eating, all the healthcare professionals and her carer were made aware of the needs to reduce her 'comfort food' intake. Her lack of understanding about the importance of 'good' nutrition required the development of an individualised health action plan, which included encouragement and support, simplifying the communication styles and provision of other activities including pastoral visits and occupational therapy. However, in spite of all intervention the patient continued to eat and gain weight.

Outcomes

The surgical debridement of the patient's haematoma resulting from a neurofibromatosis lesion completely healed following 11 weeks of treatment with Silvercel dressings. Through use of the dressing, further surgical procedures were avoided which could have been potentially life-threatening due to the patient's recurrent PE and the failure of current anticoagulant therapy to reduce her risk of developing intravenous clots. Led by the TVNs, inter-trust collaboration and a close working relationship between the primary and secondary care multidisciplinary teams enabled seamless transitions of care.

Discussion

Clinical governance acknowledges that clinical decision making is a risk-management process (Department of Health, 1998). Risk taking is part of the TVN's role, creating precedents, monitoring and evaluating practice, weighing up the risks and benefits to the patients when implementing care (Scanlon and Dowsett, 2003). The TVNs involved in this case were aware that the effects of longer-term use of silver impregnated dressing currently is 'unknown'

in complex wound management. With an awareness that this patient could not be fully mobilised and therefore rehabilitated until her wound healed, the TVAs 'risky' the continuous use of an antimicrobial alginate dressing as an adjunct to the overall management of a wound that was already heavily colonised and, due to urinary and faecal incontinence, had the potential to continue to be at continual risk of further bacterial critical colonisation.

This patient's wound had a high bacterial bioburden and high levels of exudate and Silvercel was selected and used as the primary dressing until the wound healed. The authors, however, do not advocate the long-term use of silver dressings and would advise caution when using such a dressing.

Conclusion

This case study illustrates the benefits of close collaboration between secondary and primary care in the management of a patient with such a rare condition. It highlights the necessity for holistic assessment and a multidisciplinary approach to wound management. It also illustrates the benefits of using an antimicrobial dressing as an adjunct to the overall management of a wound with a heavy bacterial bio-burden. By reducing the risk of wound infection, the clinicians successfully avoided further surgical procedure, which could have been potentially life-threatening for the patient and which could have led to further complications.

This patient had a long history of complex poly-pathologies and complications associated with NFI and as a result had many differing needs as a result of her comorbidities. The close effective collaborative working relationship between TVAs within the primary and secondary care settings enabled seamless transition of care and led to improved clinical outcomes. **WUK**

References

Department of Health (1998) *A First Class Service. Quality in the New NHS*. DoH, London

Department of Health (2001) *Valuing People: A New Strategy for Learning Disability for the 21st Century*. DoH, London

Department of Health (2004a) *Choosing Health*. DoH, London

Department of Health (2004b) *The NHS Knowledge and Skills Framework (NHS KSF) and Development Review Process*. DoH, London

Lansdown A (2004) Nutrition 2: a vital consideration in the management of skin wounds. *Br J Nurs* 13(20): 1199–210

Mahoney C (1999) Aids to help district nurses boost patient nutrition and wound care. *Nurs Times* 9(32): 49–51

Meaume S, Van de Looverbosch D, Heyman H, Romanelli M, Ciangherotti A, Charpin S (2003) A study to compare a new self-adherent soft silicone dressings with a self-adherent polymer dressing in stage 2 pressure ulcers. *Ostomy Wound Manage* 49(9): 44–5

Neno R, Neno M (2006) Promoting a healthy diet for older people in the community. *Nurs Stand* 20(29): 59–65

Riccardi V M, Eichner JE (1986) *Neurofibromatosis: Phenotype, Natural History and Pathogenesis*. Johns Hopkins University Press, Baltimore, MD: 89–101, 162–8, 235–48

Russell L (2001) The importance of nutritional status in wound healing. *Br J Nurs* 10(6): S42–49

Scanlon L, Dowsett C (2003) Clinical governance in the control of wound infection and odour. In: White R ed. *The Silver Book* Quay books, Salisbury: 69–79

Schrimsher GW, Billingsley RL, Slopis JM et al (2003) Visual-spatial performance deficits in children with neurofibromatosis type 1. *Am J Med Genet* 120A: 326–30

Thomas S (2003) Atraumatic dressings. *World Wide Wounds* 1: 9

White R, Cooper RA and Kingsley A (2001) Wound Colonization and infection: the role of antimicrobials and guidelines in management. *Br J Nurs* 10(9): 563–78

White R, Cooper RA (2003) The use of topical antimicrobials in wound bioburden control. In: White R, ed. *The Silver Book*. Quay books, MA Healthcare Ltd, Salisbury: 46–59

Williams L, Leaper D (2000) Nutrition and Wound Healing. *Clinical Nutrition Update* 1(5): 3–5