CASE REPORT

Isolated lower limb hypoplasia secondary to congenital varicella syndrome: a rare occurrence and management of its complications

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SUMMARY

Isolated lower limb hypoplasia is a rare consequence of maternal congenital varicella syndrome (CVS). The hypoplastic limb is susceptible to multiple injuries, including fractures, especially if there is associated muscle weakness and lack of sensation. We describe a unique index case of a woman aged 26 years with a background of CVS who presented with a distal femur fracture following a fall onto her insensate, hypoplastic right leg. This report highlights the complexities involved in the diagnosis and management of fractures in patients with an anaesthetic limb, and in particular describes limb amputation as a successful treatment modality for distal femur fractures.

At the time of the fall, the patient initially presented to the ED of a neighbouring hospital. However, due to lack of obvious injury on plain films and the patient remaining largely pain-free, she was discharged home with advice to come back if the situation changed.

She reattended our ED 2 weeks later with increased swelling and erythema to her right knee. Lower limb examination demonstrated no external abrasions or cuts but complete loss of sensation over her right leg. She displayed only a small amount of hip extension and no active hip flexion. She had absent deep tendon reflexes on her right leg with normal reflexes on her other three limbs.

BACKGROUND

Congenital varicella syndrome (CVS) is a rare disorder affecting 2% of babies born to mothers who contract the varicella zoster virus during their first two trimesters of pregnancy.¹ The condition was first described by LaForet and Lynch in 1947,² and since then ~130 cases have been described in the literature.¹ Even though the classical symptoms of CVS such as dermatomal skin lesions, neurological defects and hypoplasia of isolated digits have been documented, little exists in the way of isolated lower limb hypoplasia. We describe a rare distal femur fracture of an isolated and complete right hypoplastic limb secondary to CVS in a patient aged 26 years. The case highlights the difficulties in decision-making and the important multidisciplinary steps taken to manage the fracture and with an above knee amputation (AKA).

CASE PRESENTATION

A woman aged 26 years with a background of CVS presented to our emergency department (ED) 2 weeks following a fall onto her right knee. She had isolated but complete congenital hypoplasia of her right limb with widespread lack of sensation and muscle weakness of her entire right lower limb secondary to CVS. This resulted in little control of movement below the hip in her right leg. At the age of 3, the patient underwent a trans-tibial amputation and mobilised with non-weight-bearing crutches as she was unable to tolerate a limb prosthesis. Moreover, her anaesthetic leg meant she also sustained multiple painless burns on her right lower limb, for which she has needed extensive grafting.

INVESTIGATIONS

Initial radiographs obtained during her first presentation to ED (figures 1–3) following the fall showed no obvious fractures; although marked osteoporosis of the distal femur was noted.

Plain film radiographs taken at our ED 2 weeks following the fall (figures 4 and 5) showed a comminuted distal femur fracture and femoral hypoplasia.

TREATMENT

In view of her complex medical history, the options of definitive treatment were discussed with the patient. She described her right lower limb as a 'useless lump of meat' and was keen to avoid long, drawn out treatments that may include a significant chance of complications. Options discussed were: non-operative management using a cast, operative management using open reduction and fixation with a plate or nail or AKA.^{3 4} A stabilised knee replacement was not discussed. After discussion with the patient and advice from a rehabilitation medicine consultant, it was decided that an AKA would be the best management of her injury; however, with little expectation that she would use a prosthesis following surgery in view of her hip muscle weakness and never having learnt to walk on two legs. Hip disarticulation is an alternative, but is a much more major and rarely performed operation, and causes much greater alteration of body image than an AKA. Complete loss of the femur would impair her sitting balance and would make it even more difficult to suspend a cosmetic prosthesis should she wish to use one in a wheelchair. Pre-amputation prosthesis was not considered in view of the dead weight from the affected leg.

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Figure 1 Initial radiographs following fall. No obvious fractures visible. Marked osteoporosis and femoral hypoplasia noted.

Figure 2 Initial radiographs following fall. No obvious fractures visible. Marked osteoporosis and femoral hypoplasia noted.

The advantages of reduced weight when mobilising, reduced risk of repeated femoral/periprosthetic fractures, the risk of further soft tissue necrosis/infections (especially in view of her skin grafts) were weighed up against disadvantages of a longer, more major surgery and the psychological/social effects of an AKA.^{5 6} In her opinion, her right lower limb was a hindrance and she was keen to proceed with any treatment that would reduce the risk of further fractures secondary to falls. She felt that non-weight-bearing mobilisation with a shortened stump would help with her balance and reduce the load on her when using her crutches. She was fully aware of the extra risks associated with an AKA, including bleeding risk, infection and longer time under anaesthesia. However, she preferred one operation compared with the possibility of multiple surgeries due to recurrent falls and fractures.

Procedure

With the patient's consent, the senior author (WS) planned and marked the patient's leg with deep consideration on the best thigh length to leave for potential future prosthesis use (figures 6 and 7).

Consideration was also paid to the skin grafts/scars and the potential for blood vessel disruption. The procedure was performed with two equal anterior and posterior flaps as is routine for AKA.

OUTCOME AND FOLLOW-UP

At her 1-week check-up, her right leg stump (figure 8) was healing well with no obvious signs of postoperative

Figure 3 Initial radiographs following fall. No obvious fractures visible. Marked osteoporosis and femoral hypoplasia noted.

complications. She developed a small blister inferior to the suture line which resolved spontaneously. Radiographs (figure 9) showed that the fractured distal femur had been removed completely. She continued to mobilise well on crutches postsurgery. The patient felt well in herself and is awaiting follow-up for consideration of a prosthesis.

Figure 4 Radiographs 2 weeks following fall showing a comminuted distal femur fracture and femoral hypoplasia.

Figure 5 Radiographs 2 weeks following fall showing a comminuted distal femur fracture and femoral hypoplasia.

In our artificial limb service, there are multiple criteria for consideration of a prosthesis. For this patient, these include:

1. Fitness of the patient's body to stand and hop without a prosthesis, including her cognitive ability to learn to mobilise with a prosthesis on her stump.
2. The ability of her stump to tolerate socket. Loss of sensation and bony prominences on the end of the stump with minimal muscle padding are risks for skin breakdown.
3. Control of her stump which depends on muscle power, length of stump and consistency of the stump; for example, firm muscle compared with redundant fat.

In this patient's case, there was no active hip flexion or control, and she had spent much of her life hopping on

Figure 6 Preoperative images of the marked leg prior to surgery.

Figure 7 Preoperative images of the marked leg prior to surgery.

Figure 8 One week postoperative wound check.

crutches. In view of this, a hip disarticulation prosthesis may be more appropriate for her in future than a trans-femoral prosthesis.

DISCUSSION

There is a 2% risk of developing CVS in fetuses that are exposed to the varicella zoster virus in the first two trimesters of

Figure 9 Postoperative radiograph of the right femur.

pregnancy.¹ Of those 2%, only 44% go on to have limb hypoplasia and other limb abnormalities. Of these, we have been unable to find any cases of isolated limb hypoplasia. This patient presented to us with isolated right lower limb hypoplasia and neurological disturbance to her right leg. She had multiple falls and skin injuries to this leg and had yet to find a treatment option that provided her with a definitive solution to her repeated injuries. The literature and common practice suggest that knee replacement would be a viable option,³ with such an injury. However, this case brings with it unique challenges; of ensuring that the underlying osteoporotic bone was strong enough to hold a replacement knee; of trying to operate on an area of skin which is extensively damaged due to burns and of the psychological challenges facing a young woman aged 26 years at the prospect of further surgery on her limb.

The case highlights the difficulties in decision-making with someone who has a fracture with CVS. However, there is no literature or guidance on what options would be suitable for someone in this situation, although management of fractures in people with spina bifida and polio is similar. Positives that can be drawn from this case include the involvement of a multidisciplinary approach to management options. It was clear that there were multiple complex issues outside of surgery that needed to be addressed before definitive treatment was decided.

Learning points

- Isolated lower limb hypoplasia is a rare occurrence in patients with congenital varicella syndrome (CVS) with little evidence or guidelines in the literature about treatment options available for distal femur fractures of the hypoplastic limb.
- Above knee amputation is an appropriate treatment modality for distal femur fractures in patients with CVS.
- Complex cases involving amputation need a multidisciplinary approach to management options.

Contributors SM, foundation year 2 doctor, was involved in the initial clerking and management of patient in the emergency department, acquisition of patient consent and images in theatre, assisted in above knee amputation with senior consultant, case write up and guarantor of the report. WS is the senior trauma and orthopaedic consultant in charge of the case, and its operative and management decisions. SK is the rehabilitation consultant who kindly saw the patient in preamputation clinic with advice on prosthesis and psychosocial support. AA is the trauma and orthopaedic consultant who assisted in case write up, editing and guidance on submission of case report.

Competing interests None declared.

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