CASE REPORT

Hanging on by a thread: a rare case of secondary pseudoainhum

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Accepted 16 January 2016

SUMMARY

Pseudoainhum is a rare and poorly characterised phenomenon involving autoamputation of a digit associated with an identifiable or related disease process. We describe the case of a 19-year-old man with long-standing neurodisability, sensorineural hearing loss and severe keratoderma, presenting with painless rapidly-progressing autoamputation of his left little toe. Genetic analysis thus far has been inconclusive. The toe appeared to be hanging on by a thread, with no clinical signs of infection. Radiographs showed lysis with complete resorption of the proximal phalanx. Routine blood tests found an isolated neutropenia with normal C reactive protein and biochemistry. We report an unusual case of a young adult with a history of chronic neurological and dermatological problems presenting with secondary pseudoainhum and provide a valuable addition to the limited literature describing this rare condition.

BACKGROUND

Constricting bands on the digits were first described almost a century ago¹ and have been characterised by authors as ainhum, pseudoainhum, dactylosylis spontanea and congenital bands, with some inconsistency in the literature over nomenclature. A recent review clarified the distinction²: ainhum refers to idiopathic autoamputation of a digit as a result of a constricting band or groove, usually in the toes of dark-skinned patients; pseudoainhum describes autoamputation as a result of an associated disease process. Pseudoainhum can be further classified as primary, whereby it presents at birth/in early life, or secondary when presenting later in life. Contrary to ainhum, pseudoainhum is not associated with particular ethnic backgrounds and can occur anywhere in the body.

The aetiology of pseudoainhum is poorly understood, and infection, constitutional racial differences, mechanical effects associated with hyperkeratosis and vascular changes have been implicated.² It is thought that histological changes in pseudoainhum resemble those in ainhum, which initially presents with gross thickening of the stratum corneum, and calcification of the ducts and the sweat glands. At a more advanced stage, there is chronic inflammation below the hyperkeratotic epithelium. As the fissure deepens due to this fibrotic reaction of collagen with a hyperkeratotic epidermis, tendons, vessels and nerves are compressed with eventual absorption of the bone by pressure. The latter stages are generally thought to be sterile and avascular,³ although Browne argued

that it represented an 'infection-healing-fibrosis-ulceration' cycle.⁴ There is an expanding body of case reports describing distinct cases of pseudoainhum, but no larger studies have looked at a cohort of patients with the condition or charted the histology or radiology associated with the condition. Pseudoainhum has been identified in rare heritable syndromes including Kindler syndrome⁵ and Vohwinkel syndrome,⁶ suggesting a genetic component.

CASE PRESENTATION

We present the case of a 19-year-old man with a background of severe learning difficulties, developmental delay, sensorineural hearing loss, hypoplastic corpus callosum, arthrogryphosis with previous hamstring release and neutropenia in infancy (thought to have resolved). Genetic analysis thus far has been inconclusive, although he shares some features of arthrogryphosis, renal dysfunction and cholestasis (ARC) syndrome.⁷

He was referred to the Foot and Ankle Orthopaedic Department at our institution by a dermatologist who was treating his severe palmar-plantar keratoderma and fungal toe infections, with a 2-week history of autoamputation of his left little toe. His parents described thinning around the base of the toe occurring in the past with limited spontaneous resolution prior to this episode. This was painless and he was systemically well throughout.

On examination, there was a constriction ring around the base of the left little toe, with the toe attached to the foot by a minimal amount of soft tissue, as if it were hanging 'by a thread' (figure 1). Severe changes associated with keratoderma were noted on the plantar aspect of the foot (figure 2). A constriction band was emerging around the base of the left fourth toe also, while the right foot and

Figure 1 The dorsal aspect of the patient's left foot.

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To cite: Arkush L, De Silva B, Gordon D. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2015-213854

Figure 2 The plantar aspect of the patient's left foot.

hands were uninvolved. There were no signs of infection on the right foot or toe.

INVESTIGATIONS

Radiographs showed osteolysis with complete resorption of the proximal phalanx (figure 3). Routine biochemistry was normal and C reactive protein was <1 mg/L. Full blood count revealed an isolated neutropenia (Neut $0.71 \times 10^9/L$).

TREATMENT

The clinical scenario was thought to be consistent with secondary pseudoainhum, associated with the patient's underlying medical condition. The patient was offered surgical termination of the fifth toe if he became symptomatic from pain or inconvenienced by the toe catching during daily activities, but this was declined in the absence of signs of infection.

DISCUSSION

The previous literature suggests that pseudoainhum may be associated with hyperkeratotic changes.¹ This is consistent with our presentation of a patient with severe plantar keratoderma and previous case reports suggesting that a common pathological process may be implicated.^{8–11} The association of this presentation with a recurrence of neutropenia may shed further light on the aetiology of this condition, suggesting a possible immune component, although this may reflect a coincidental finding.

Figure 3 Dorsoplantar and oblique radiographs of the patient's left foot.

The short natural history of this patient's pseudoainhum is unusual. True ainhum has been characterised as being a slowly progressive condition⁴ and most case reports suggest pseudoainhum similarly follows a long course,^{12 13} although there is one description in the literature of a 1-week history of pseudoainhum in a patient suffering from acute psoriasis.¹⁴ This is of clinical significance, as by the time he presented, autoamputation was impending, giving limited opportunity for preventative therapy.

Pseudoainhum has been described as being an infrequent complication in the autosomal recessive keratodermas¹¹ and has been reported to occur in Vohwinkel syndrome, also known as keratoderma hereditaria mutilans.⁹ Likewise, pseudoainhum has been associated with a novel mutation in a Connexin gene in a patient with palmoplantar keratoderma and sensorineural hearing loss.¹⁰ Although genetic analysis is yet to identify a distinct genetic abnormality in our patient, there appears to be a common theme running through the literature concerning patients with genetic abnormalities developing pseudoainhum—they frequently present with sensorineural hearing loss and palmoplantar keratoderma.

Constrictions bands of the toes, we believe, should be treated based on symptoms rather than appearance, where surgical intervention should be considered if there is pain, infection or a functional limitation.

We present a case of pseudoainhum in a patient with a complex medical history with features of ARC syndrome. Constricting bands eventually leading to autoamputation of digits in adults is thought to be an under-reported finding in the literature,¹⁵ and early recognition in patients with complex conditions such as this could avert such an outcome. While the association of pseudoainhum with keratoderma and with sensorineural hearing loss has been previously reported in the literature, further evidence is required to confirm the association of these rare presentations and to delineate whether there may be a shared aetiology.

Learning points

- ▶ Pseudoainhum involves autoamputation of a digit associated with an identifiable or related disease process.
- ▶ The natural history of autoamputation can vary from weeks to years.
- ▶ While some patients are pain-free, symptoms can involve pain, infection or the digit catching during daily activities.
- ▶ Pseudoainhum is associated with genetic conditions involving keratoderma and sensorineural hearing loss.
- ▶ Management should be guided by the patient's symptoms, and often conservative management is advised.

Contributors LA drafted the manuscript. DG and BDS provided valuable feedback, advice and corrections on the manuscript, and further insights into the patient's history and presentation.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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