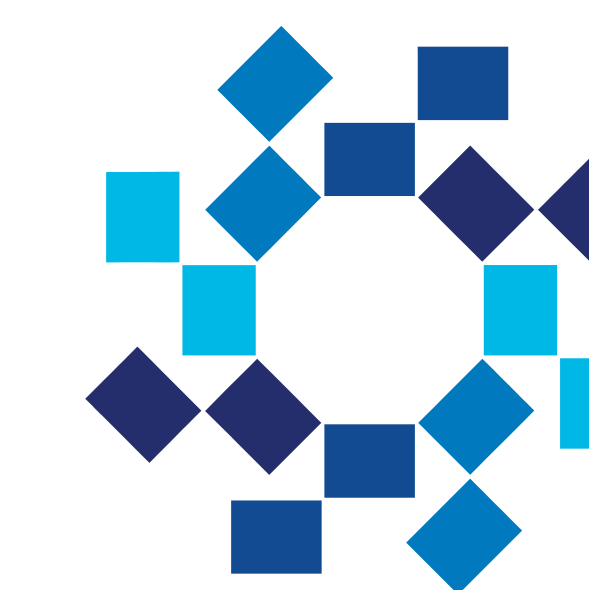


Recurrent Infections in the Setting of Proteus Syndrome Affecting Unilateral Lower Extremity: A Case Report

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STATEMENT OF PURPOSE

1. To present a rare case of Proteus Syndrome with recurrent lower extremity infections

PROTEUS SYNDROME

- Proteus syndrome (PS) is a rare and highly variable developmental genetic disorder characterized by somatic tissue hyperplasia.
- PS is caused by AKT1 gene mosaic mutations, regularly associated with growth regulation.
- The incidence of Proteus syndrome is less than 1/1,000,000 live births.
- Individuals affected are born without any noticeable symptoms, although with age, irregular, disproportionate, and asymmetric tissue overgrowth occurs.
- Physical symptoms of tissue overgrowth range from mild to life-threatening, including osseous, connective tissue, and vascular malformations.
- Vascular malformations can lead to increase risk in deep vein thrombosis (DVT)



Figure 1: Clinical presentation of Proteus syndrome foot with hypertrophic osseous and connective tissue over-growth

CASE STUDY

- Male patient in his thirties presented with a history of Proteus syndrome diagnosed in childhood.
- Patient's chief complaint of acute onset erythema and warmth to the left hallux. Plantar distal hallux wound measuring 1.5cmx1.2cmx0.2cm is noted down to and including subcutaneous tissue.
- He had an extensive history of multiple infections to the left foot, all which have resolved without complications.
- Radiographic and advanced imaging evaluation showed prior partial hallux amputation, extensive first ray bony hyperplasia, and an osseous block to the anterior ankle, but ruled out deep abscess.
- The patient ambulates with the ankle in a plantarflexed position due to the osseous ankle block. The patient bears weight entirely to the plantar distal aspect of the hallux: correlating to the area of skin breakdown and infection. Remainder of the extremities are within normal limits.
- The patient at this time denies any surgical intervention for reconstruction of the foot.
- The area of pressure was appropriately offloaded; local wound care was performed to the area, and the patient was initiated on a course of antibiotics.



Figure 2: DP and lateral radiographic views of first ray bony hyperplasia and extensive osseous bony block

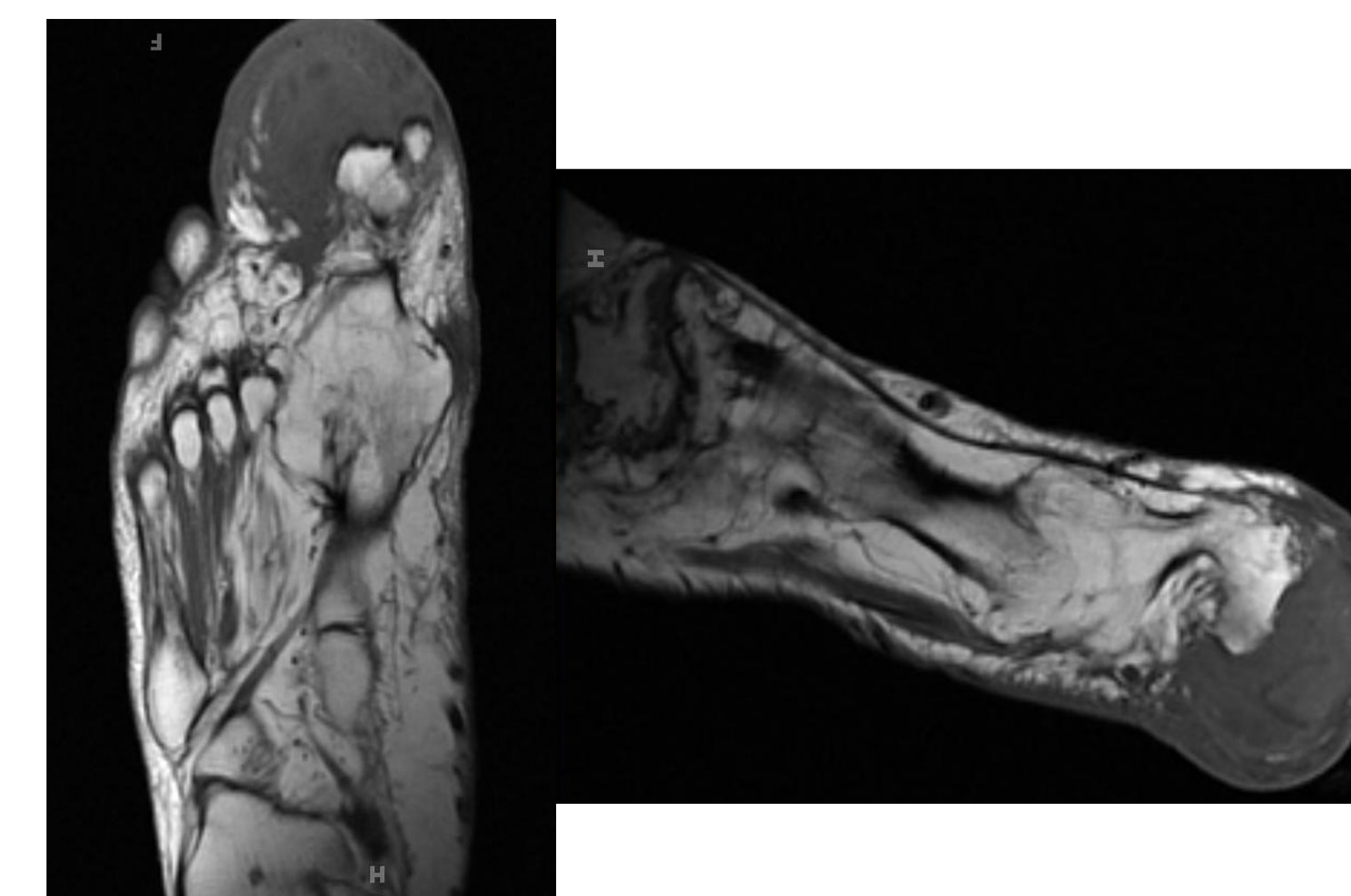


Figure 3: Advanced magnetic resonance imaging; showing no bony or soft tissue infectious processes

ANALYSIS & DISCUSSION

- Upon completion of an antibiotic course, the patient was instructed to remain non-weightbearing at all times to ensure adequate wound healing. Patient was given a rolling knee walker as well as crutches.
- A complete reconstruction of the affected foot was discussed, which the patient opted against at this time.
- Frequent podiatric follow-up and vigilance is imperative.
- The rare and highly variable nature of Proteus syndrome makes diagnosis challenging and treatment complex.
- Special care must be taken to treat the various musculoskeletal malformations and recurrent infectious processes related to this disease.
- Special care towards DVT prophylaxis in surgical situations is a must.
- Current literature supports symptom-specific treatment, including podiatric specific interventions.

REFERENCES

1. Jamis-Dow, C. A., Turner, J., Biesecker, L. G., & Choyke, P. L. (2004). Radiologic Manifestations of Proteus Syndrome. *RadioGraphics*, 24(4), 1051-1068. doi:10.1148/rg.244035726
2. Keppler-Noreuil, K. M., Lozier, J. N., Sapp, J. C., & Biesecker, L. G. (2017). Characterization of thrombosis in patients with Proteus syndrome. *American Journal of Medical Genetics Part A*, 173(9), 2359-2365. doi:10.1002/ajmg.a.38311
3. Proteus syndrome - Genetics Home Reference. (n.d.). Retrieved October, 2017, from <https://ghr.nlm.nih.gov/condition/proteus-syndrome#resources>