中文題目：Gorham-Stout 病引起之右脛骨及腓骨復發性病理性骨折

英文題目：Gorham-Stout disease with recurrent pathologic fracture over the right tibia and fibula

作　　者：蔡宗翰[1], 蘇祐立[2]

服務單位：[1] 高雄長庚紀念醫院內科部, [2] 高雄長庚紀念醫院血液腫瘤科

Introduction

Gorham-Stout disease (GSD, also known as Vanishing bone disease) is a rare disease of unclear etiology. It is pathologically characterized by endothelial-lined vessel proliferation in bone and progressive osteolytic destruction due to osteoclast activation. Clinical symptoms include bone pain, swelling and deformity. In some cases disease cause disruption of lymphatic tissues, resulting in chylothorax, pericardial effusion and chyloperitoneum. Treatment options include surgery, radiotherapy and pharmaceuticals including bisphosphonates and mTOR inhibitors. However, there are no gold standard for optimal medical therapy. We presented a 55-year-old female with history of right proximal fibula excision due to Gorham-Stout disease with recurrent right lateral knee pain and swelling despite pain-free status for nearly 5 years.

Case presentation

A 55-year-old female presented with recurrent right lateral knee pain since September 2020. The pain is exacerbated by walking and poorly relieved by medication. She had similar symptoms in 2015 and diagnosed of right proximal fibula Gorham-Stout disease via excisional biopsy. She lost follow-up and remained pain-free during ambulation for 4 years. However, she experienced worsening right knee pain for several months. The plain film of the right knee showed erosion and scalloping of the right distal fibula and pathologic fracture of the proximal tibia. Excision of the osteolytic lesion and ORIF was done. Immunostaining of biopsy specimens consist of increase capillaries and small lymphatics, confirming recurrent Gorham-Stout disease. The patient had received sirolimus and radiotherapy since September 2021.

Conclusion

The diagnosis of GSD is challenging due to its rarity and variable presentation. Other etiologies of osteolytic lesions (malignancy, infection, autoimmune disease or

metabolic osteodystrophy) should be carefully excluded. There is no standard therapy for GSD due to lack of large-scale studies. Sirolimus (mTOR inhibitor) as a target for angiogenesis and cellular proliferation, along with good safety profile and efficacy through case reports, constitutes a favorable pharmaceutical option. For rapidly progressing disease or refractory cases, local management such as surgery and radiotherapy should be considered.