Tania Amin1, Brook Adams2, Jeanette Kraft3 and Valentina Leone1

1Department of Paediatric Rheumatology, 2Department of Paediatric Radiology, Leeds Children Hospital, Leeds, UK

**Background:** Primary hypertrophic osteoarthropathy (PHOA1) is a rare genetic condition caused by mutation of the hydroxyprostaglandin dehydrogenase (HDPG) gene leading to prostaglandin excess, with digital clubbing, acro-osteolysis, periostitis, arthropathy and skin changes including pachyderma, hyperhidrosis and excessive facial furrowing [1]. Radiological findings previously described include plain radiographic changes with irregular periostal proliferation, cortical thickening of long bones and acro-osteolysis. Bone scintigraphy identifies characteristic changes of symmetrically increased periarticular linear uptake of the tracer along the distal ends of long bones [2]. Joint US examination has been reported in one case only revealing synovitis and inflammation. Management options described in case reports include long-term NSAIDs, bisphosphonates, DMARDs, infliximab and arthroscopic synovectomy with variable symptomatic relief, and no improvement in established plain radiographic changes. Although the disease has been described as self-limiting, significant pain and disability is reported.

**Aims:** We describe two unrelated children with clinical and radiological correlation, including the use of MRI, which has not previously been reported.

**Methods:** Case 1: a 15-year-old male patient, previously diagnosed with PHOA1 in 2007 (homozygous mutation of the HDPG gene), was referred to paediatric rheumatology in 2012 with a year of progressive bilateral knee and leg pain. On examination he showed bony swelling of ankles and knees with restriction and marked finger clubbing. His older sister was also severely affected. Case 2: a 3-year-old female patient with consanguineous parents, the only affected child of six siblings, was diagnosed with homozygous mutation of the HDPG gene following a referral at 2 years of age with delayed walking. There was a history of surgical ligation for a large PDA at 4 months of age. On examination there was marked bilateral knee swelling with fixed flexion deformities and finger clubbing.

**Results:** Case 1: MRI and plain radiographs were taken synchronously. Hand radiographs demonstrated soft tissue swelling and fingertip expansion but no acro-osteolysis. Otherwise, plain radiography showed normal bony appearances. MRI of knees and ankles showed effusions with synovial enhancement, marked periostitis affecting the patella and distal tibia, and tenosynovitis at the ankle with minimal change following treatment with a prolonged course of regular long-acting NSAIDs and a short course of steroids. Case 2: a skeletal survey demonstrated long bone diaphyseal periosteal reactions and acro-osteolysis affecting both hands and feet, consistent with hypertrophic osteoarthropathy. MRI revealed expanded, thickened and enhancing cortices suggesting periostitis. The surrounding muscle showed patchy oedema in keeping with an inflammatory condition.

**Conclusion:** This is the first report of MRI changes in PHOA1. We plan to treat this disabling condition with pamidronate using serial MRI to monitor treatment response.

**Disclosure statement:** The authors have declared no conflicts of interest.

**Reference**