Hyperphosphatemic tumoural calcinosis

Sahana Shetty,[1] Nitin Kapoor,[1] Sarah Mathai,[2] Thomas Vizhalil Paul[1]

[1]Department of Endocrinology, Christian Medical College (CMC), Vellore, Tamil Nadu, India
[2]Department of Child Health-1, Christian Medical College, Vellore, Tamil Nadu, India

Correspondence to
Professor Thomas Vizhalil Paul, thomasvpaul@yahoo.com

Accepted 9 January 2016

DESCRIPTION

A 13-year-old girl presented with pain and progressive swelling over both hips of 2 years duration along with restriction of movements at the hips. There was no history of trauma, back ache, fever, renal calculi, polyuria, fractures and involvement of other joints. There was no similar illness in other family members. On examination, there were swellings of more than 20 cm over both hips with painful restriction of movements. The rest of the examination was unremarkable. The patient's blood biochemistry revealed a high-phosphorus level of 6 mg/dL (N: 2.5–5). The rest of the bone biochemistry was normal. The patient's pelvis X-ray showed features of tumoural calcinosis (TC) on both sides (figure 1). A diagnosis of TC with hyperphosphatemia was performed. The patient was initiated on a low-phosphate diet and phosphate binding agent sevelamer.

TC is a rare syndrome characterised by calcium deposition in soft tissues adjacent to the joints. It usually starts in adolescence as painless, firm, tumour-like masses around the joints resulting in restriction of joint function.[1] TC can be of two types: normophosphatemic and hyperphosphatemic. Mutations in the gene encoding *FGF23*, *GALNT3* and *KL* have been implicated in hyperphosphatemic familial TC.[2]

Chronic kidney disease has been described to be associated with secondary TC. The typical radiographic appearance of amorphous, cystic and multilobulated calcification located in a periarticular distribution along the extensor surfaces along with the biochemical hallmark of hyperphosphatemia caused by increased renal absorption of phosphate in the absence of renal dysfunction is diagnostic of TC. Genetic analysis may further delineate the underlying genetic mutation responsible for the metabolic defect.

TC can be diagnosed easily if the characteristic radiological findings as described above are present. However, for subtle cases, the diagnosis can be established with the help of isotope bone scan, CT or MRI. Histopathology showing multiple cystic spaces with large geographic areas of calcification surrounded by palisaded histiocytes and numerous foreign body type giant cells is confirmatory in doubtful cases.

Management is multidisciplinary, including dietary restriction of phosphate and phosphate lowering agents such as aluminium hydroxide and sevelamer. Surgical excision is indicated when lesions are large with limitation of movements or where there is an associated infection.[1]

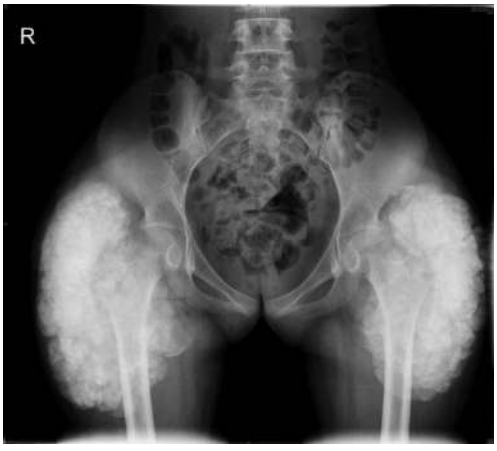

Figure 1 Pelvis X-ray showing bilateral tumoural calcinosis.

Learning points

- Tumoural calcinosis (TC) is a rare syndrome characterised by calcium deposition in soft tissue regions adjacent to the joints, and has its onset in adolescence as painless, firm, tumour-like masses around the joints resulting in restriction of joint function.
- Management of hyperphosphatemic TC includes dietary restriction of phosphate and phosphate lowering agents such as aluminium hydroxide and sevelamer.
- Surgical excision is indicated when lesions are large with limitation of movements or where there is an associated infection.

Contributors SS, NK and SM wrote the manuscript; SS, NK, SM and TVP reviewed the manuscript and gave final approval.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

1 Fathi I, Sakr M. Review of tumoral calcinosis: a rare clinico-pathological entity. *World J Clin Cases* 2014;2:409–14.
2 Rafaelsen S, Johansson S, Ræder H, *et al*. Long-term clinical outcome and phenotypic variability in hyperphosphatemic familial tumoral calcinosis and hyperphosphatemic hyperostosis syndrome caused by a novel GALNT3 mutation; case report and review of the literature. *BMC Genet* 2014;15:98.

To cite: Shetty S, Kapoor N, Mathai S, *et al*. *BMJ Case Rep* Published online: [*please include* Day Month Year] doi:10.1136/bcr-2015-213537