Hyperphosphatemic tumoural calcinosis

Sahana Shetty,1 Nitin Kapoor,1 Sarah Mathai,2 Thomas Vizhalil Paul1

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

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.

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REFERENCES
