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Untreated congenital hypothyroidism in limping child: A rare case with epiphyseal dysgenesis and vertebral anomaly

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Abstract

Hypothyroidism associated with skeletal abnormalities are uncommon these days due to early neonatal screening. In low demanding low socio-economic group in later age of the life, may present with advanced epiphyseal dysgenesis or dysplasia associated with abnormal vertebrae. This many of time confuses with other diagnosis if thyroid profile is not detect. Later in age these children may presented with limping gait along with restricted motion. Primary survey of radiographic images often misdiagnosed as perthes or Slipped capital femoral epiphyseal (SCFE) or multiple epiphyseal dysplasia if history is not clear. Therefore, it is wise to raise suspicion on thyroid profile derangement of the child presenting with painless limping gait with restricted motions, along with atypical dysplastic hip joint or bullet shape vertebrae. The following case is reported because there was no associated dwarfism, delayed milestone and apparently no familial achondroplastic trait.

Keywords: Hypothyroidism, Epiphyseal dysgenesis, SCFE, Perthes, Achondroplastic

Introduction

In our case Eleven-year-old girl with painless limping presented ^[1]. On initial survey perthes and SCFE was kept in the mind. Radiography ^[2] revealed Unilateral epiphyseal dysgenesis of the proximal femur, associated acetabular and vertebral changes. Earlier diagnosis of hypothyroidism is difficult to make before three year of age which leads to undiagnosed and untreated skeletal deformities. Hypothyroid screening is usually done in neonatal age group. Especially in developing countries, it is wise to suspect early these patients presentation, but relatively difficult if no other features of hypothyroid is been seen such as delayed milestones or short stature. Miss diagnosis is common in such cases in low socio-economic groups where that leads to untreated skeletal deformities ^[6].

Case Report

11-year-old girl presented with limping gait in the orthopaedic department. Which was progressive in nature in last 5 months, associated with on and off pain radiating from left hip to thigh and back. She was first born child vaccinated without an eventful birth history. Patient was up to normal developmental milestone with adequate stature. No endocrinological screening was done in her neonatal periods. On examination she was having restricted abduction and internal rotational movements in left hip without any scarring or history of previous infection. She was having trendelenburg gait associated with shortening in affected leg. We suspected perthes or resolved TB hip. Routine investigations were normal. X ray of hip (FIG.1) with knee joint and LS spine (FIG.2) was obtained. Showing stippled epiphyseal dysgenesis of the head of the femur ^[2] along with the unusual shape of the vertebra in lower thoracic region. MRI (FIG.3) was done for confirmation of the diagnosis. Thyroid profile was revealed to be TSH = 5.74 uIU/ml and T3 = 2.83nmol/L. T4= 127 nmol/L. Hypothyroid picture was established and treated accordingly. Surgical treatment with osteotomy is planned in later settings.

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Fig 1: Left hip showing epiphyseal dysgenesis of femoral head with acetabular changes



Fig 2: Bullet shape vertebra in lower thoracic region

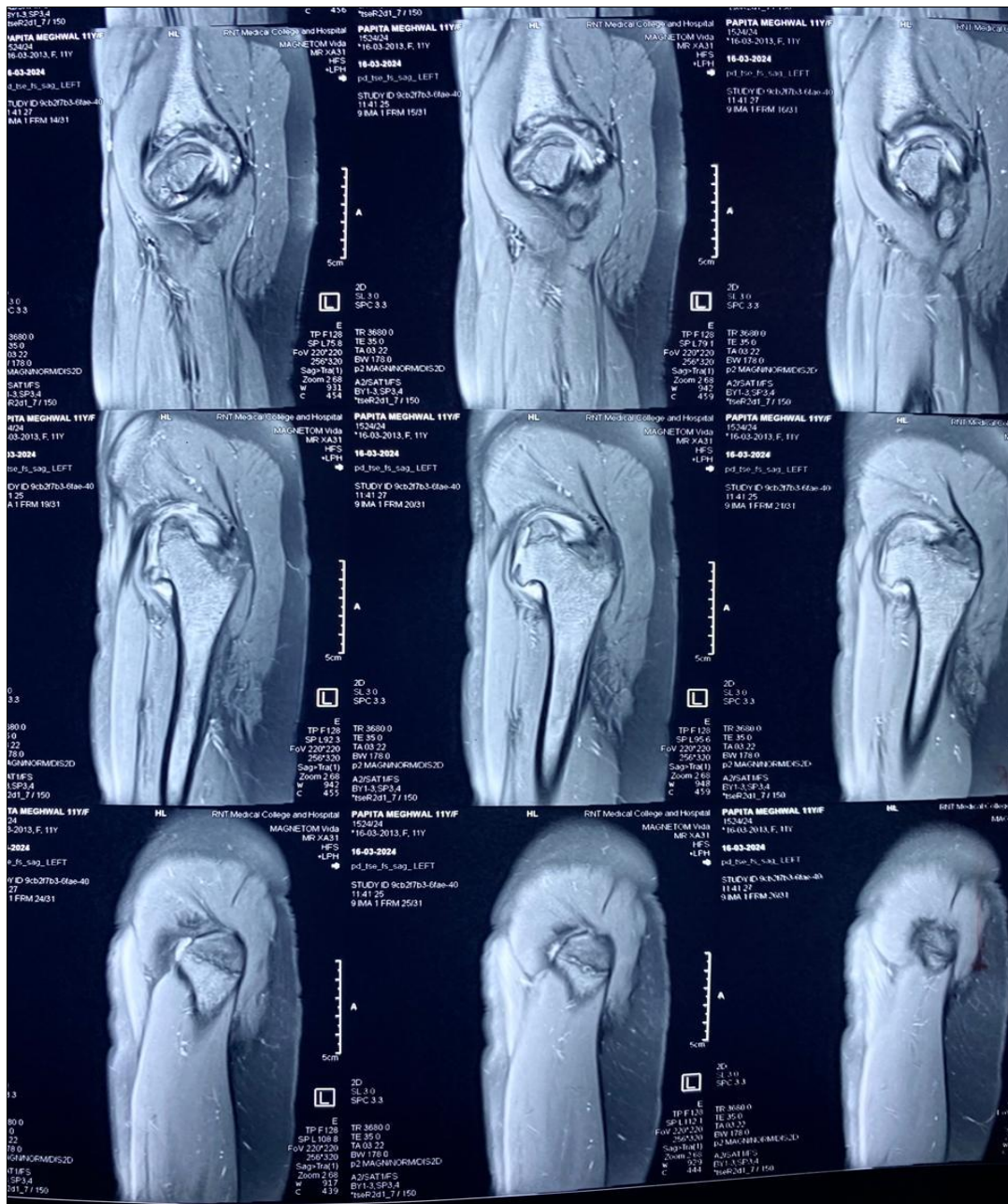


Fig 3: MRI of left hip showing irregular contour of femoral head

Discussions

Thyroid hormone integrates their roles with growth development and maturation of the skeleton by regulating chondrocyte proliferation, mineralisation and angiogenesis [5]. In medical literature hypothyroidism associated epiphyseal ossification changes was noticed by 1990. Disappearance of epiphyseal dysgenesis was noted after one year of starting the therapy. This particular case was not associated with additional finding such as short stature or hearing loss or with short arms and delayed mile stone. Such pictures itself makes the diagnosis difficult to establish on initial survey [6]. The Hypothyroid patients with these findings are typically older.

Conclusion

These days in modern clinical practice it is rare to see untreated hypothyroidism with skeletal abnormalities because of early screening and awareness. It is important to diagnose early and early treatment of hypothyroidism for the clinicians. But this is challenging to diagnose as in this oligosymptomatic case. Specially in developing countries where neonatal screening [4] is not well established in low socioeconomic regions. Misdiagnosis or late diagnosis is very common in such presentations. Multiple joint Plain x ray films along with radiological spine screening is wise to obtain in the initial survey. Congenital spondylodysplasia [3] and perthes are in differentials if proper history and laboratory markers are not available. Hence radiological and endocrinological discussion is wise to be done especially if the patient belongs to the developing world.

Review of Literature

Chaudhary N, Sharda S *et al* (2011) [6] Reportd a case of untreated hypothyroidism with spondyloepiphyseal dysplasia along with bilateral hip joint changes in neglected case

Consent

Written inform consent was obtained from patient's parents as patient was minor, for publication of this case report and accompanying images.

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