CASE REPORT

Spondyloepiphyseal dysplasia tarda with progressive arthropathy associated with subcapsular cataract

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SUMMARY

Spondyloepiphyseal dysplasia tarda with progressive arthropathy is a form of inherited skeletal dysplasia involving the axial skeleton along with swelling and deformities of the peripheral joints that mimics juvenile rheumatoid arthritis. We report a case of a 14-year-old school boy who presented with dwarfism, spine deformity, ocular anomaly and peripheral arthropathy with characteristic radiological changes in the vertebrae and multiple other joints.

BACKGROUND

Spondyloepiphyseal dysplasia tarda with progressive arthropathy (SED-TA) also called progressive pseudorheumatoid arthropathy of childhood (PPAC) or progressive pseudorheumatoid dysplasia (PPD) is a form of osteochondrodysplasia, inherited usually in an autosomal recessive pattern.¹ The first case was described in 1980 and 1982.² It is an extremely rare disorder with a global prevalence of only 1/1 000 000. About two-third of reported cases were from Arabia³ and very limited number of cases have been reported from India.⁴ Although SED-TA manifests between 3 and 6 years of age,⁵ diagnosis is often delayed. However, our patient presented with stiffness and deformity of multiple joints at an early age. He also had ocular anomaly, which was an extremely rare association with PPD. Perhaps, we are the first to report the association of a subcapsular cataract with PPD.

CASE PRESENTATION

A 14-year-old boy was admitted with a history of progressive swelling, stiffness and deformity in the wrist and fingers of both hands. It started first with mild low back pain and pain in both hip joints since 6 years of age. Gradually he developed stiffness of hips with difficulty in walking. He also developed swelling and pain in both knee joints along with difficulty in flexing them. Since past 2 years he developed gradual swelling and pain of small joints of both hands which progressively resulted in stiffness and deformity of the fingers. There was no history suggestive of any joint inflammation, fever or rash. He had received analgesic occasionally from his family physician for pain relief but did not receive any steroid preparation. He had uncomplicated birth history, and normal intelligence with good academic performance. There was no history of any consanguineous marriage in the family. His family members including his two elder brothers had no similar symptoms.

He had a short stature with a height of 132 cm (vertex to pubis 60 cm and pubis to floor 72 cm) and arms span 152 cm. He had increased thoracic kyphosis and lumbar lordosis. His systemic examination was normal. Both hips had reduced abduction and lateral rotation and both knees had restricted flexion associated with mild pain on movement. Interphalangeal joints of fingers were minimally painful, swollen with fixed flexion deformity (figure 1). However, he did not have any restricted movement at the cervical and lumbar spine. He had normal audiometric results in both ears. His unaided visual acuity was 6/9 and 6/24 in the left and right eye, respectively. There was left-sided subcapsular cataract.

INVESTIGATIONS

The patient had a normal complete blood count. Erythrocyte sedimentation rate (ESR) was 28 mm and the rheumatoid factor was 16.6 IU/mL. He had normal thyroid profile, calcium, phosphate, normal liver and renal function tests. X-ray of the elbow and hand showed epiphyseal enlargement of all metacarpals, proximal and distal interphalangeal joints as well as all bones forming elbow joints, with varying degrees of osteopenia (figure 2). Lateral X-ray of the dorsolumbar spine revealed classical feature of platyspondyly with superior and inferior bony humps over vertebral bodies (figure 3). Anteroposterior X-ray view of the hip joints revealed bilateral flattened and widened epiphyses of the femoral head (figure 4). So, clinical features and investigations point to a diagnosis of sSED-TA with subcapsular cataract.

Figure 1 Flexion deformity and swelling of the interphalangeal joints of both hands.

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Figure 2 Hand X-ray showing epiphyseal enlargement of the metacarpophalangeal and interphalangeal joints with juxta-articular osteopenia.

DIFFERENTIAL DIAGNOSIS

- ▶ Juvenile rheumatoid arthritis (JIA)
- ▶ SEDT-PA
- ▶ Morquio syndrome

TREATMENT

The patient was first prescribed non-steroidal anti-inflammatory agents considering JIA but he did not respond to them. He was discharged with calcium supplements and advised physiotherapy of joints.

DISCUSSION

Spondyloepiphyseal dysplasia (SED) is a rare disorder of bone growth and articular cartilage involving spine (spondylo) and epiphyses of long bones causing disproportionate short stature. SED can be broadly classified as (1) SED congenita and (2) SED tarda.⁶ SED congenita is inherited as an autosomal dominant pattern and presents at birth with clinical features of skeletal abnormalities, cleft palate, visual and hearing defects. SED tarda can be inherited either as X linked, autosomal recessive or dominant form. SEDT-PA is a variant of SEDT which is characterised by the involvement of axial joints, swelling and deformities of small peripheral joints.³

It results from a mutation in WISP3 gene located on the long arm of chromosome 6.⁵ WISP3 gene help in the development of skeleton, nerve fibres and blood vessels by regulating cellular proliferation and differentiation and has vital regulatory effect on insuline-like growth factor 1 (IGF-1) signalling pathways. In the SEDT-PA mutant, WISP3 gene fails to regulate IGF-1 resulting in an uninhibited hypertrophy and terminal differentiation of stable articular chondrocytes.⁷ This leads to a short stature along with arthralgia and deformity in small joints of the hand.

Figure 3 Lateral X-ray of the dorsolumbar spine showing osteopenia, generalised platyspondyly with bony humps over vertebral bodies.

Figure 4 Anteroposterior X-ray of the pelvis showing flattened and enlarged epiphyses of the femoral head.

Characteristic radiological changes are seen in vertebrae associated with epiphyseal enlargement of metacarpophalangeal and interphalangeal joints. Unfortunately, this genetic mutation study could not be performed in our case, as this facility is not available in our institution.

SED-T-PA is frequently misdiagnosed as JIA since onset of SED-T-PA occur during early childhood and characterised by pain, swelling and stiffness of several joints especially hands, wrist and elbow. However, SED-T-PA lacks inflammatory changes in the peripheral joints. In the present case too an initial diagnosis of JIA was considered but ESR was normal and imaging did not reveal any feature of joint destruction. In many previous case reports too JIA has been considered a major differential for PPD.^{8,9}

Sine PPD is a form of type II collagenopathy, and type II collagen is present in hyaline cartilage, nucleus pulposus and vitreous humour of the eye, defect in collagen type II may lead to ocular anomalies.³ However, most of the case reports have found association of myopia, cataract and retinal detachment with SED congenita.¹⁰ One case report has also shown association of corneal opacity with SED tarda.¹¹ Our patient was found to have left-sided subcapsular cataract, which is an extremely rare association with SED-T-PA.

Learning points

- ▶ Spondyloepiphyseal dysplasia tarda with progressive arthropathy is an inherited autosomal recessive disorder, involving the axial skeleton as well as small peripheral joints.
- ▶ It mimics juvenile rheumatoid arthritis in clinical presentation.
- ▶ It results from mutation in WISP3 gene resulting in defects in collagen type II.
- ▶ It may be associated with extraskeletal manifestations such as cataract.

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Competing interests None.

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