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CASE REPORT

Familial pseudotail, scoliosis and synpolydactyly syndrome

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Abstract
Purpose This case series describes a novel condition characterized by familial pseudotail associated with scoliosis, and synpolydactyly that has not been previously reported in literature.
Methods The authors present three siblings and one cousin from the same family living in the northern region of the Arabian Peninsula. All cases presented with pseudotail, scoliosis, and complex synpolydactyly. The authors demonstrated complete clinical and radiological descriptions in addition the detailed performed surgeries.
Results The histopathological result of the resected pseudotail specimens revealed bony lesion covered with thick fibrous tissue and evidence of mature adipocytes within trabecular spaces.
Conclusions The described cases represent a novel condition that has not been previously reported in the literature. Familial pseudotail scoliosis synpolydactyly syndrome is a newly recognized form of familial pseudotail.

Keywords Familial · Pseudotail · Scoliosis · Synpolydactyly · Syndrome

Introduction

The etiology of the congenital sacral abnormalities is still not well understood; however, maternal diabetes was considered as a primary cause since as 16 % of the cases were born to diabetic mothers, on the other hand, other authors believed that exposure to chemicals like acetone was likely to be the cause [1]. Yet other authors reported that a familial genetic mutation was responsible for those abnormalities [2].

Caudal appendages are uncommon congenital malformations; they present as either a true tail or a pseudotail [3]. The true tail is usually rare which is remnant of the embryonic tail that contains adipose tissue, striated muscle, blood vessels, and nerves, and covered by normal skin [4, 5].

Human tail embryology is poorly understood, but usually results from the disturbance of caudal regression during the sixth week of embryonic life [6].

In the presented case series, the authors demonstrate the clinical and radiological description as well as the surgical management of a novel condition of three siblings and one cousin from the same family living in the northern region of Arabian Peninsula. The condition is characterized by familial pseudotail, scoliosis and synpolydactyly that have not been previously reported in literature.

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Materials and methods

Case 1

RA is a 12-year-old female from the northern region of the Arabian Peninsula who was born with bilateral synpolydactyly in both hands and syndactyly in both feet. She was delivered full term, normal vaginal uncomplicated pregnancy for consanguineous marriage. Her parents have no history of any medical disease.

At the age of four, she had several surgical procedures to correct her hands synpolydactyly. A year later, her parents noticed that she had a deformed back. Scoliosis screening series was done; she was found to have a scoliosis curve of around 20°. At the age of 12, her scoliosis curve progressed to reach 65°, at that time she was referred to our institution.

Her chief complaints were scoliosis, synpolydactyly type SD 2 of both hands, syndactyly of both feet and a hard mass at the end of her tailbone that caused pain on sitting. Family history reveled that she has two younger siblings with the same condition; a 10-year-old sister and 4-year-old brother.

Physical examination of her back showed left thoraco-lumbar curve, moderate hump and right proximal thoracic compensatory curve with positive bending test for scoliosis and her neurological examination was normal. There was a skin dimple with thick and pigmented overlaying subcutaneous hard bony prominence that was felt at the lower end of the tailbone. This pseudotail showed signs of chronic pressure around its tip, without pressure sore or hair tuft (Fig. 1).

Physical examination of her extremities showed synpolydactyly of both hands with multiple old surgical scares; she has syndactyly of both feet in the second and third toes were sharing skin web whereas the other toes were well separated.

Scoliosis series X-ray showed pelvic obliquity and left thoraco-lumbar curve with its apex at T12 and Cobb's angle was 90° that was measured from T5 to L4. In addition to right proximal compensatory thoracic curve with apex at T3 and Cobb's angle 40° that was measured from T1 to T5 (Fig. 2). Computed tomography (CT) scan of the lumbo-sacral spine showed transverse sacrum, retroversion of coccyx; the coccyx was pointed backward and up in lordotic orientation instead of normal kyphosis with an angle of around 90°, forming pseudotail like scorpion tail (Fig. 3). The magnetic resonance imaging (MRI) of the pseudotail showed dark signal with no nerve tissue.

The patient was referred to a pediatric cardiologist, a neurologist, and a dysmorphologist, whom confirmed that there was no other clinical abnormality. Genetic testing and chromosomal analysis were negative for any known

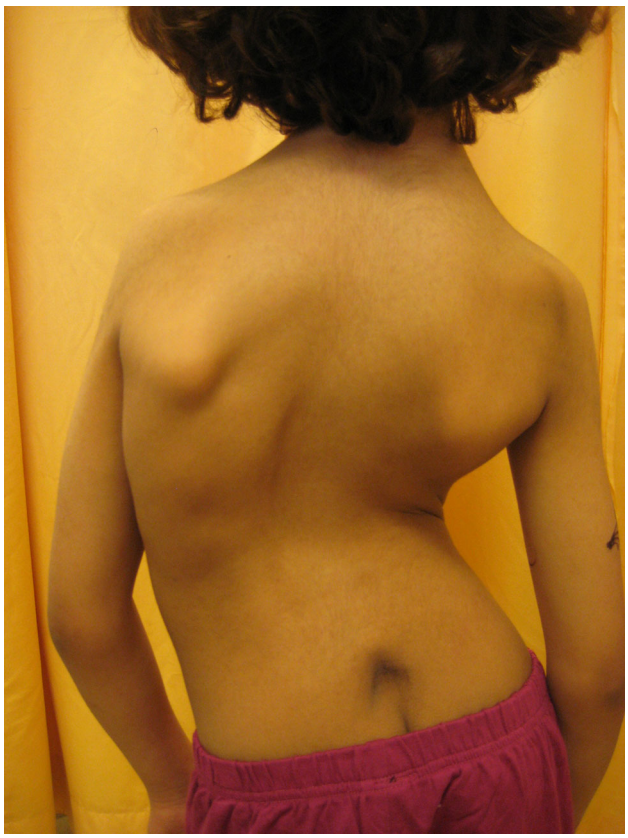

Fig. 1 Case 1 (RA): clinical photo of the back showing scoliosis and the pseudotail

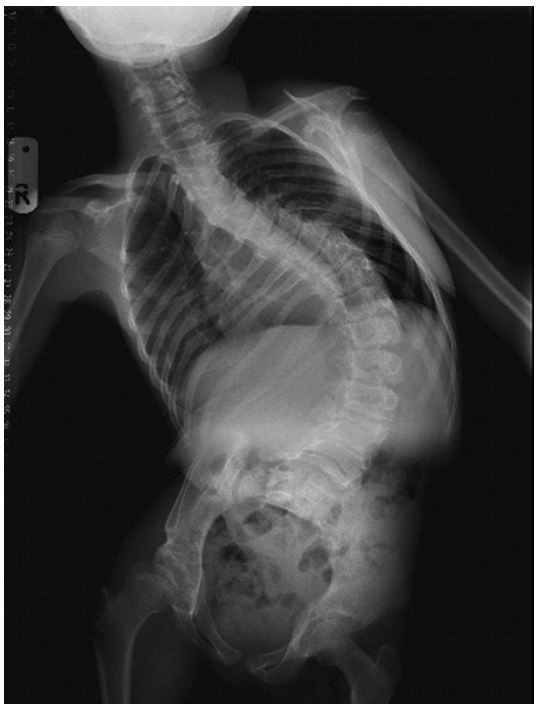

Fig. 2 Case 1 (RA): pre-operative X-ray scoliosis series; antero-posterior view showing left thoracolumbar curve scoliosis deformity

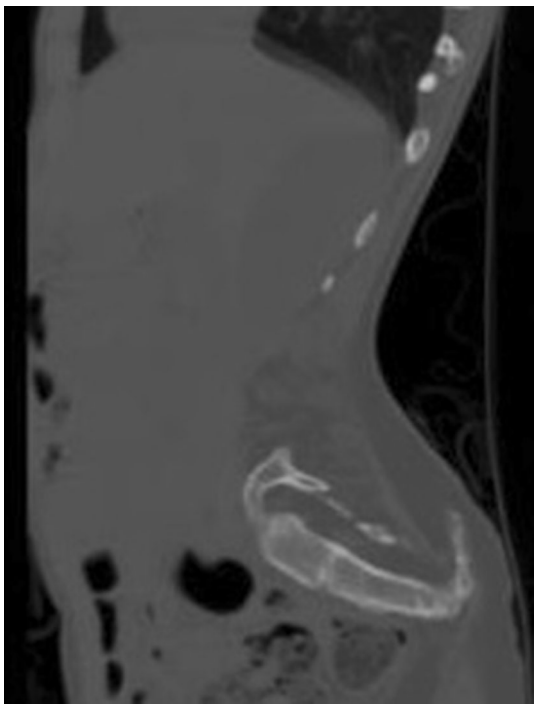

Fig. 3 Case 1 (RA): CT scan lumbosacral spine sagittal view showing the retroversion of pseudotail

genetic disorders and that case did not fit in the diagnosis of any known syndrome.

A scoliosis correction surgery with excision of the prominent pseudotail was decided by the treating surgical team. Standard posterior thoraco-lumbar spinal fusion with trans-pedicle screw fixation for scoliosis correction of the spine from T2 to the iliac bone under intra-operative neurophysiology monitoring was done. Thereafter, skin incision was extended caudally for better exploration of the lordotic coccyx, it was found to be posteriorly curved and retroverted. The retroversion angle of the pseudotail was approaching 90°, the distance between the curved part and body of the coccyx (about 2 cm) was filled with fat. The curved part of the coccyx was dissected followed by excision of the pseudotail just proximal to the curved part of the coccyx.

The post-operative course was uneventful; the patient was followed up monthly for 1 year then annually for 3 years with satisfactory outcomes (Fig. 4a, b).

Case 2

ARA is a 10-year-old female; the younger sister of case 1 (RA). She presented with synpolydactyly of her hands and feet, mild scoliosis and pseudotail. She had similar perinatal history to her older sister (case 1). She was evaluated by pediatric dysmorphologist and a geneticist and they did not find her condition fit at any known syndrome.

Her back examination showed mild right thoracic curve with positive bending test for scoliosis. A hard bony prominence was felt subcutaneously at the lower end of the tailbone, with mild skin pigmentations and thickening around its tip without a pressure sore or hair tuft (Fig. 5).

She had synpolydactyly type SD 2 in her both hands. Both feet showed proximal syndactyly between the first and the second toes and complete syndactyly of the second and third toes, while the other toes were well separated.

A scoliosis series X-ray showed mild right thoracic curve; Cobb's angle was about 12° as measured from T5 to T12 (Fig. 8). A CT of the lumbo-sacral spine demonstrated a defect of the posterior element of the lower sacral vertebrae, moreover the sacrum was occupying a horizontal position; the coccyx was curved backward and upward in retroverted angle about 90° to form hook-shaped pseudotail abutting the subcutaneous tissue (Fig. 6). The spine MRI showed no tethered cord, no myelocoele nor any abnormality in the spinal cord. The space between the pseudotail and the coccyx was filled with soft tissue and had the same signal intensity of fat.

Due to the presence of only mild scoliotic deformity, no surgical correction was offered. Pseudotail excision was recommended to relieve her sitting pain. However, the parents opted to defer the procedure until such time when scoliosis correction surgery was needed. The patient was followed up every 6 months, and she showed no further progression at her most recent visit.

Case 3

ANA is a 4-year-male; the younger brother of RA and ARA (case 1 and 2) who presented with the same triad; pseudotail, scoliosis, and synpolydactyly. His perinatal history was uneventful. Synpolydactyly was noticed shortly after delivery, and he was medically evaluated, but his condition did not fit any known syndrome.

Upon arrival to our institution, his physical exam did not show any neurological deficit. His back examination showed scoliosis with positive bending test, in addition to the skin pigmentation at the lower part of the tailbone, in addition to a hard bony mass under the skin (Fig. 7).

A scoliosis series X-ray confirmed the left curve scoliosis; Cobb's angle was 40° which was measured from T7 to L2. A lumbo-sacral spine CT scan showed spine segmentation abnormality and it detailed the anatomy of the pseudotail. The tip of the coccyx was pointed backward to be a pseudotail directly under the skin (Fig. 8). The lumbo-sacral spine MRI demonstrated tethered cord with protruded pseudotail that showed dark signal with no nerve tissue.

Surgery for releasing the tethered cord and excision of the pseudotail was recommended and brace was advised

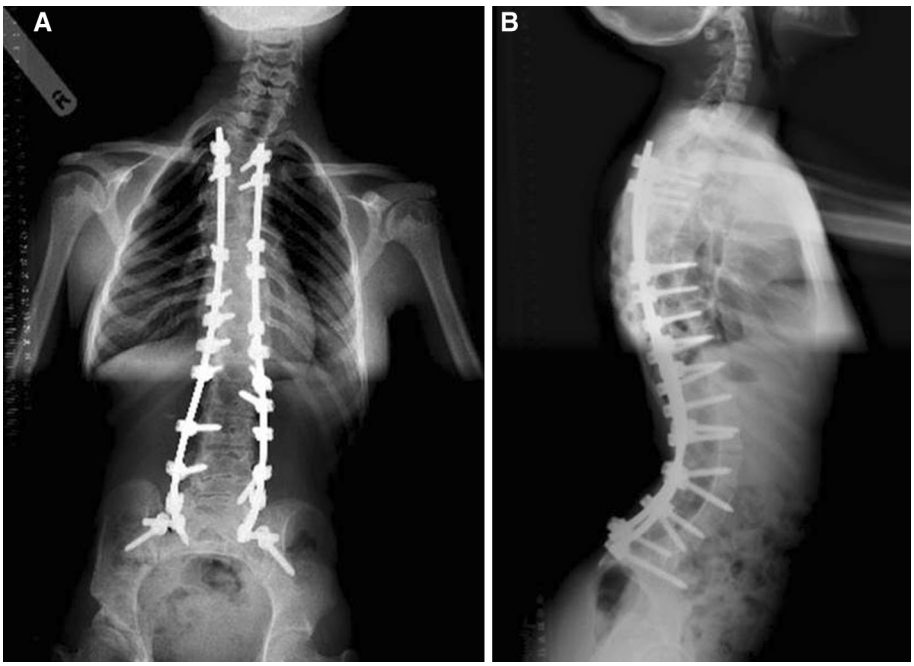

Fig. 4 Case 1 (RA): post-operative X-ray scoliosis series. **a** Antero-posterior view shows correction of scoliosis. **b** Lateral view shows correction of scoliosis and pseudotail excision

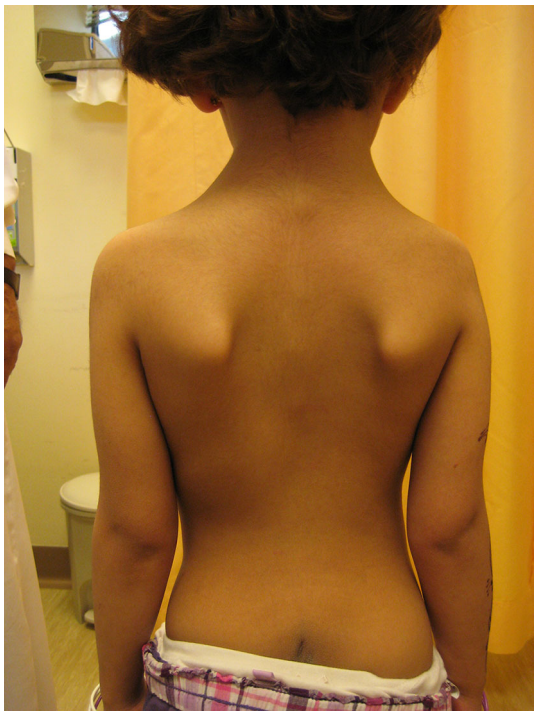

Fig. 5 Case 2 (ARA): clinical photo of the back showing mild scoliosis and pseudotail

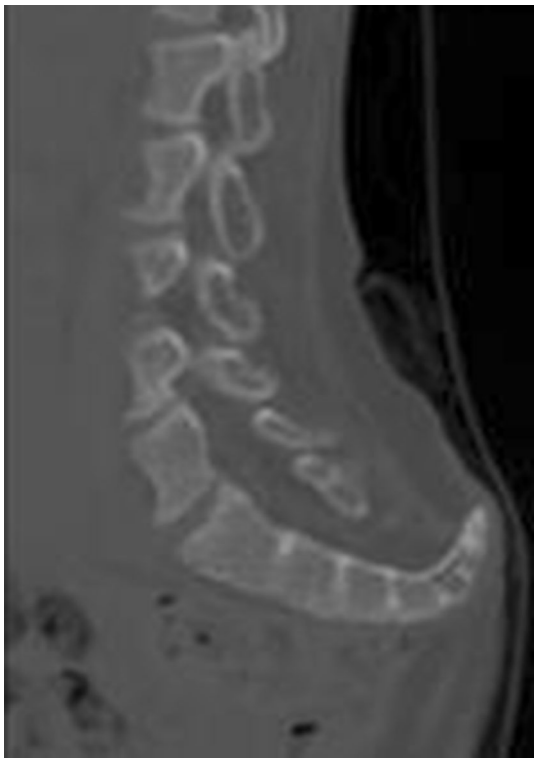

Fig. 6 Case 2 (ARA): CT scan lumbosacral spine sagittal view showing the pseudotail

for scoliosis. Three months later, the patient underwent surgery for release of the tethered cord. Surgical incision was extended distally to expose the pseudotail where the curved part of the coccyx was excised subperiosteally.

Patient was followed up every 6 months with an X-ray scoliosis series, but his scoliosis curve showed progression reaching a Cobb's angle of 78° (Fig. 9a). Consequently, scoliosis correction using expandable prosthesis was recommended. The previous surgical incision was utilized to insert the lower foundation of the expandable prosthesis at

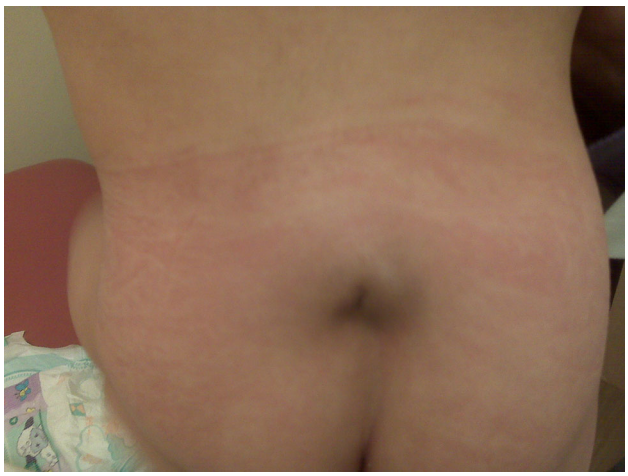

Fig. 7 Case 3 (ANA): clinical photo of the back showing the pseudotail

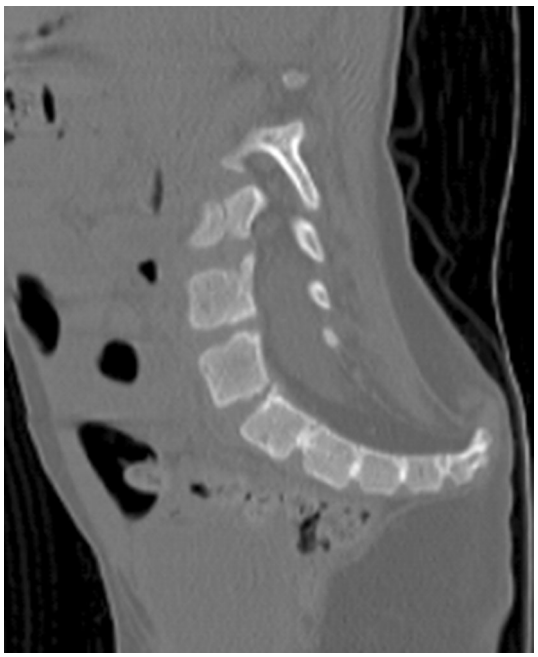

Fig. 8 Case 3 (ANA): CT Lumbosacral spine sagittal view showing pseudotail

the level of L4–L5, while the upper foundation was applied at the T3–T4 level using a separate incision. This was followed by subcutaneous insertion of the expandable prosthesis. Six months later, the patient underwent expandable prosthesis elongation procedure (Fig. 9b).

Case 4

ABA is a 12-year-old male; he is the cousin of the former described three cases. His parents have a consanguineous marriage; his father is the uncle of the previously described three cases, and his mother is the aunty of the described cases as well. The patient presented with same triad; pseudotail, scoliosis, and synpolydactyly.

His back examination showed scoliosis and pseudotail that was felt subcutaneously at the end of the tailbone (Fig. 10).

The medial four fingers of the right hand shared single web syndactyly together (mitten hand). The left hand fingers shared a common skin web that was surgically separated. The right foot showed syndactyly of the middle three toes, whereas the left foot showed third and fourth toes syndactyly.

A spine scoliosis X-ray series demonstrated moderate right curve scoliosis with a Cobb's angle of 38° that was measured from T4 to T10. A CT lumbar-sacral spine showed the details of the pseudotail, which had an upward orientation of the coccyx, in addition to a horizontal sacrum, spina bifida occulta from S2 downwards (Fig. 11). The MRI whole spine showed a tethered cord with low insertion of the conus medullaris at the level of L2–3. Syringomyelia was demonstrated in the cervical and thoracic cord from C3 to T11. The spinal cord in lumbar region showed dural ectasia without spinal dysraphism.

Surgical excision of the pseudotail was recommended for cosmetic reasons and to alleviate the pain during sitting.

The standard posterior approach was carried out to expose the tailbone; a skin incision was done over the most prominent part, followed by subcutaneous dissection until the pseudotail was fully exposed. The retroverted coccyx was excised just proximal to the retroverted part followed by skin closure. However, the release of the tethered cord was planned to be carried out if the scoliosis surgery was to be done.

Results

Both parents of the described four cases (RA, ARA, ANA and ABA) were evaluated clinically and radiologically and there was no evidence of scoliosis, syndactyly or pseudotail. They have no history of any medical illness as well. The exact relationship between the three siblings and their cousin and the consanguinity in the family is presented in family pedigree (Fig. 12).

All presented cases underwent several reconstructive and corrective surgeries for synpolydactyly in both hands.

All cases were presented with congenital scoliosis, two of them; case 1 (RA) underwent posterior scoliosis correction surgery with spinal fusion, and case 3 (ANA) underwent expandable prosthesis insertion 6 months after tethered cord release surgery. Both of them were followed up with an uneventful post-operative course. Due to the mild scoliosis curve of the other two cases ARA and ABA, surgery is not advised, but they were followed up regularly.

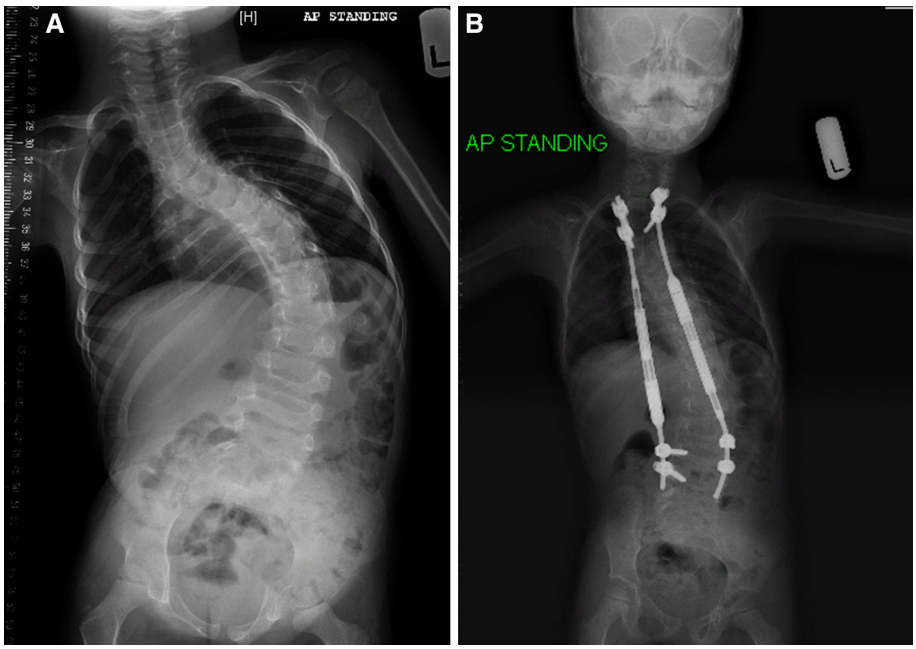

Fig. 9 Case 3 (ANA): **a** pre-operative X-ray scoliosis series antero-posterior view showing progressed scoliosis curve. **b** Post-operative X-ray scoliosis series antero-posterior view showing improved scoliosis curve after surgery

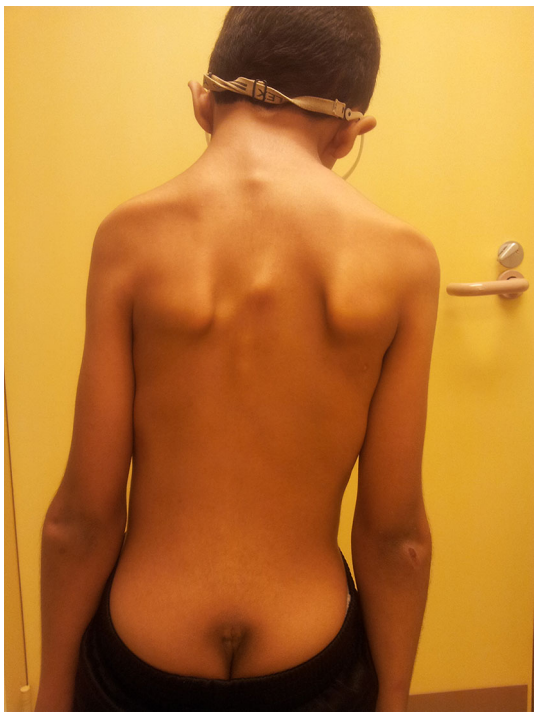

Fig. 10 Case 4 (ABA): clinical photo of the back showing scoliosis and pseudotail

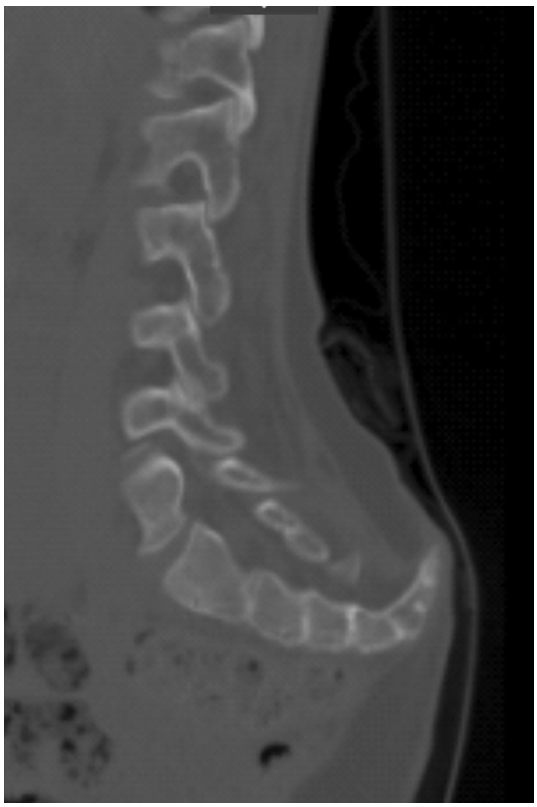

Fig. 11 Case 4 (ABA): CT lumbo-scaral spine; sagittal view showing the pseudotail

All cases have the pseudotail, which was a protrusion of the tail vertebra, with sitting associated problem and skin pigmentation over the hard bony mass. Three of the presented cases; case 1 (RA), case 3 (ANA) and case 4 (ABA) underwent excision of the pseudotail with acceptable results.

Case 2 (ARA) was advised for the excision of the pseudotail, but her parent opted to delay the procedure and to be combine with scoliosis correction surgery if needed.

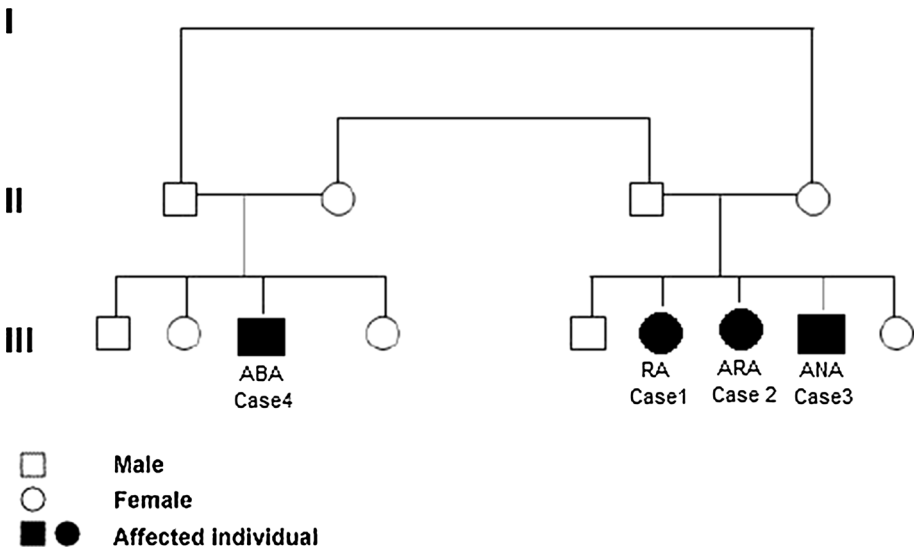

Fig. 12 Family pedigree of the studied cases

The excised pseudotail masses from case 1 (RA), case 3 (ANA), and case 4 (ABA) were sent for histopathology, which revealed bone, muscles adipose tissue and no nervous tissue was detected.

Discussion

The pseudotail is a protrusion of the lumbosacral area, which is also known as tail-like appendages usually contain bone, cartilage, notochord and elements of spinal cord [7].

There are several presentations of pseudotail; the most common one is the protrusion of the coccygeal vertebra in the lumbosacral region, and other presentations include lipomas, teratoma, chondromegaly, and glioma [3].

Lendon et al. classified sacral abnormality into three groups; complete absence of the vertebra hemivertebra, and neuronal arch deficiency. However, neither pseudotail nor sacral retroversion was included in his classification [8].

In the literature review carried out by Venkataramana et al., they found over 100 reported cases of human tail; these cases were usually associated with complex spinal abnormalities. Human tail can be associated with spina bifida, tethered cord syndrome, intra-spinal lipoma, spinal dysraphism, lipo-myelomeningocele, lumbo-sacral teratoma, terminal myelocele and coccygeal vertebra [9].

A literature review of the period between 1960 and 1997 was carried out by Samura et al., revealed 59 cases that some of them were associated with congenital dermal sinus [2]. A few cases of pseudotail were reported. Pouzet reported familial pseudotail in only one case in 1938 [6]. The association of pseudotail with scoliosis and synpolydactyly has never been reported in the literature.

In the present work, the authors report four cases with in the same family; three siblings and their cousin, who presented with the same triad; pseudotail, scoliosis, and synpolydactyly. The authors selected familial pseudotail scoliosis synpolydactyly syndrome (FPSSS) to represent this syndrome.

In the presented cases, scoliosis correction surgery was advised in a high degree scoliotic curve as in case 1 (RA) that underwent posterior scoliosis correction surgery with spinal fusion or in an ongoing progressive scoliotic curve as in case 3 (ANA) that underwent expandable prosthesis insertion 6 months after tethered cord release surgery. In cases with mild scoliotic curve as in the other two cases ARA and ABA, surgery was not advised, but they needed to be followed up regularly.

The pathogenesis of the pseudotail is not well understood, there is no set standard for the treatment of pseudotail. The surgical excision of the pseudotail was done to relief the pain upon sitting, and for better cosmetic appearance.

Sequels of pseudotail excision are not well studied in the literature, but there are few reports of sacral regeneration after sub-periosteal excision of the sacro-coccygeal vertebra after long-term follow-up [1]. Neurological deficits are not expected as there are no nerves passes through the coccyx, but pseudotail excision may interfere with the integrity of the inserted ligaments.

The described cases represent a novel condition that has not been previously reported in the literature. FPSSS is a new form of familial pseudotail.

Compliance with ethical standards

Conflict of interest None of the authors has any potential conflict of interest.

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