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Case Series

A Rare Combination of Clubfoot and Down Syndrome: A Case Series and Literature Review

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ABSTRACT

Down syndrome (DS) is commonly associated with pesplanovalgus among foot disorders due to hypotonia and joint laxity. Soft tissue contractures around the ankle and foot are implicated as the pathoanatomy in the case of Clubfoot, which is different from DS. Few cases of Clubfoot have been reported in association with DS. We report two cases of Clubfoot in association with DS, which were treated with the ponseti method and gave a good result.

Keywords: clubfoot, CTEV, case report, downs syndrome, hyperlaxity

INTRODUCTION

Down syndrome (DS) is the most common chromosomal anomaly caused by trisomy 21 and is associated with multiple anomalies.[1] Hypotonia, joint laxity, increased Body Mass Index(BMI), and ankle instability contribute to most orthopedic problems associated with DS.[1-6] Twenty percent of all DS have orthopedic problems; among them, 60-91% have pesplanovalgus.[1,3,6,7] Other orthopedic manifestations in DS are scoliosis, atlanto-axial instability, hip subluxation, slipped capital femoral epiphysis, Legg-Calve-Perthes disease, and patellar instability, and in the foot other than pesplanovalgus hallux valgus, syndactyly, clinodactyly. The pathoanatomy of Clubfoot is the talocalcaneonavicular (TCN) dislocation with soft tissue contractures around the ankle, which is strikingly different from the DS.[8,9] So, the occurrence of DS and Clubfoot is less reported.

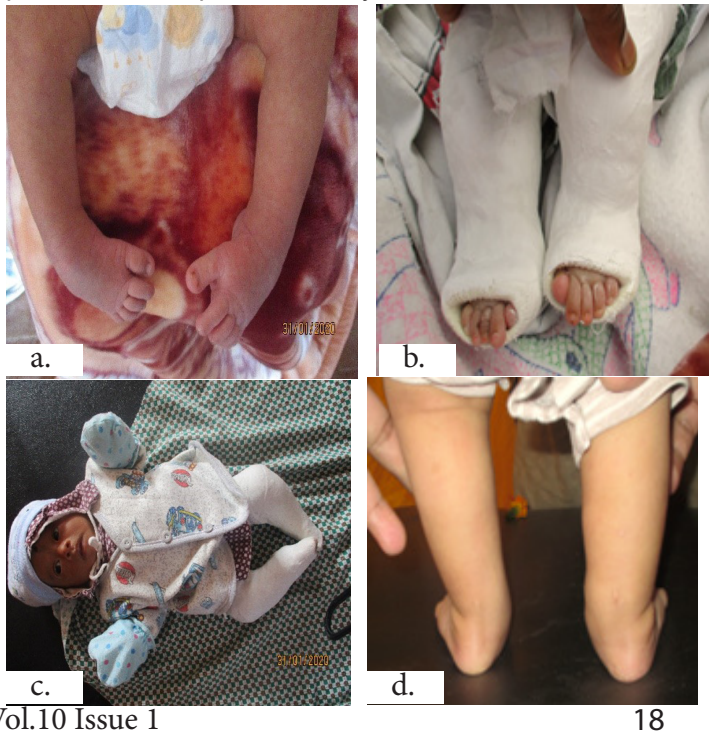
We were presented with 2 cases of Clubfoot in DS, and we are presenting them.

CASE 1

Currently, a four-year-old boy who was term delivered by cesarean section without any complication during pregnancy and postnatal period had no family history of club foot. He presented to our center at the age of 1 month with a complaint of bilateral foot deformity by his parents. (Figure 1) On clinical examination, he had typical Down syndrome-like facies, epicanthal folds with slanting palpebral fissures, and a wide gap between the first and second toes, all indicating the physical features of Down syndrome. On regular follow-up, he had a developmental delay and started walking only at the age of 2 years. Besides that, he had no other cardiac and GI defects associated with Down syndrome.

Local examination showed equinus, heel varus, forefoot adduction, and cavus deformity observed in the bilateral foot with normal spine, hip, and upper extremities. (Figure 1) His Pirani score was 5 with a hind foot score (HFS) and midfoot score (MFS) of 2.5 each and a dimegilo score of 14 on both feet during his first visit.

We started manipulation and serial casting by the ponseti method at the age of 1 month. After three ponseti casting, a Pirani score of 1 with MFS 0 and HFS 1 was achieved. We planned tendoachilles (TA) tenotomy after that. After this, his dorsiflexion at the ankle was 10 degrees, and foot abduction was 30 degrees. The final cast was applied for three more weeks, and started on a foot abduction brace (FAB) afterward. The patient was on FAB up to 22 months, and then recommended for club foot shoe for daytime after he started to walk. (Figure 1) He is on regular follow-ups at our center, and at the last follow-up, at the age of 4 years, he had 10 degrees of dorsiflexion at the ankle and had the ability to squat. The patient is ambulatory and on regular pediatric checkups for Down syndrome.



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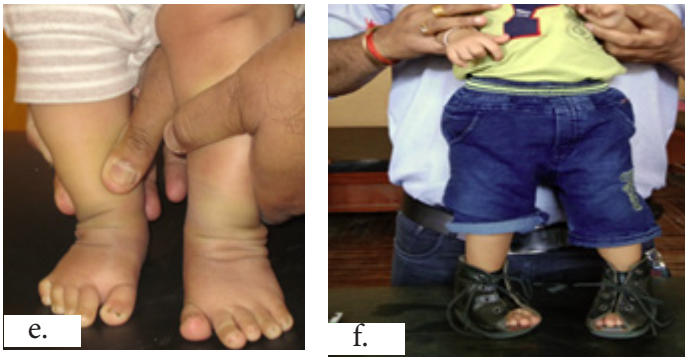
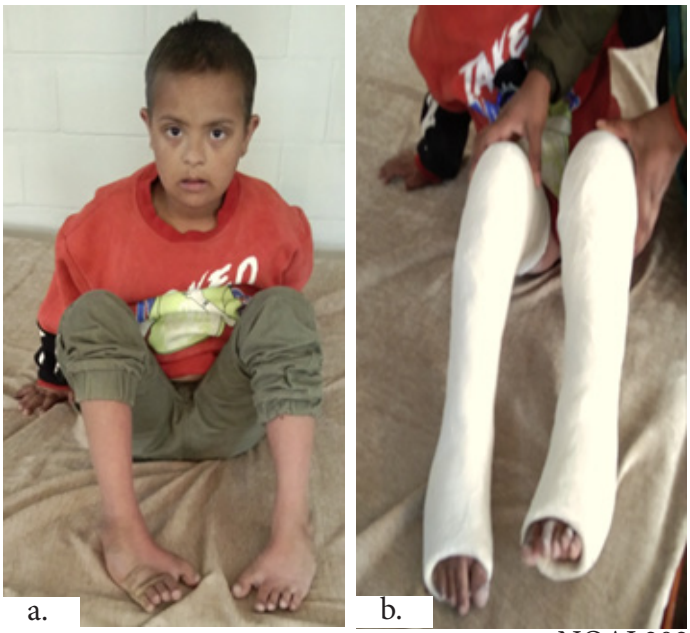


Figure 1: a. Space between 1st and 2nd toe with Clubfoot, b. After 1st ponseti cast, c. Cast before tenotomy, d. After TA tenotomy, good valgus at the ankle is present, e. Plantigrade foot, f. Case in FAB

CASE 2

Our second case is nine years boy, preterm, hospital vaginal delivery with a history of jaundice and NICU admission for it. He was presented to our center for bilateral foot deformity by his parents at the age of 18 months with already-diagnosed Down syndrome and associated atrial septal defect, patent ductus arteriosus (PDA), and hypothyroidism. He was on thyroxine for hypothyroidism. The patient was planned for serial ponseti casting but lost to follow-up for a long time and presented seven years later in our center. During those periods, he had main pulmonary artery banding with PDA ligation and band for ASD. On clinical examination, he had typical physical features of Down syndrome, having a mongloid face, epicanthal folds with upslanting palpebral fissures, flattened nasal bridge, and wide space first toe.

On examination, he had Clubfoot deformity in both feet. The spine upper extremity is normal with the hyper mobile hip joint. On a visit at the age of 9 years, his Pirani score was 4 and Dimeglio score of 12 on bilateral feet and managed to drop to 2(MFS 0 HFS 2) after ponseti casting five times. He was then planned for TA lengthening with posterior release. Due to the unavailability of cardiac backup in our center we referred the case for operative management at



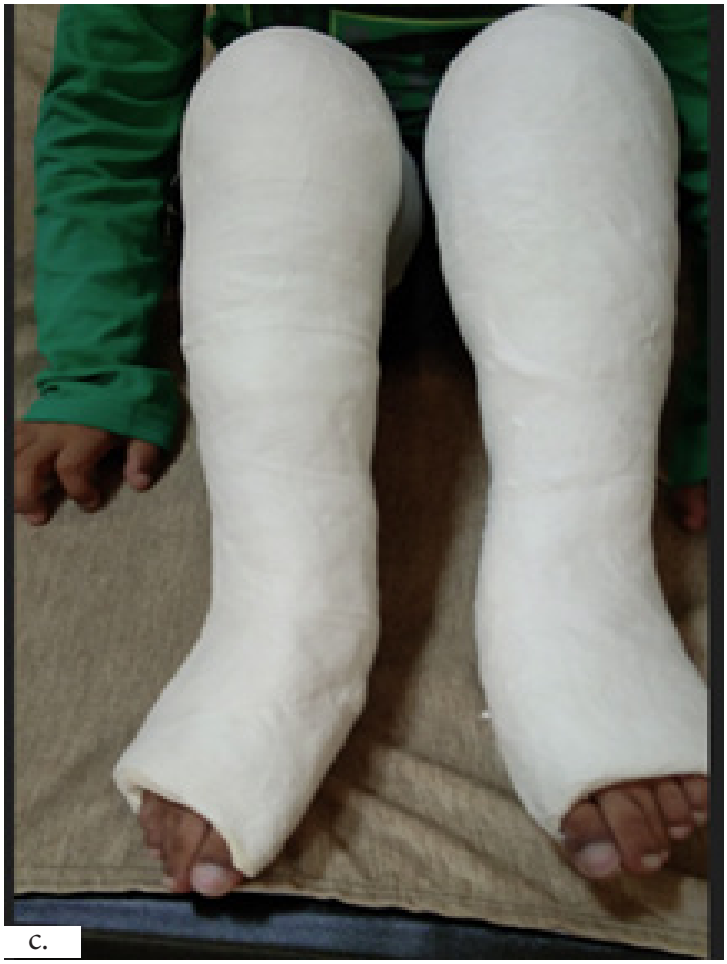


Figure 2: a. Space between 1st and 2nd toe with bilateral Clubfoot, b. After 1st ponseti cast, c. 5th Cast

the center with cardiac backup. The patient has been lost to follow-up since then.

DISCUSSION

Clubfoot is one of the most common congenital orthopedic anomalies, with incidence reported at 1 in 1000 live births.[8-10] Clubfoot can be classified as idiopathic, postural, syndromic, or neurologic.[8,9] Different syndromes have been implicated in Clubfoot, but its incidence in DS is less reported. Syndromic and neuromuscular Clubfoot tends to be more rigid and difficult to treat compared with idiopathic Clubfoot.[8,9] The pathoanatomy of Clubfoot is the talocalcaneonavicular (TCN) dislocation with soft tissue contractures around the ankle, which is strikingly different from the DS.[8,9] Both our patients had bilateral clubfeet, which were supple and had low pirani scores at presentation, contrary to the other resistant syndromic Clubfoot.

DS is the most common chromosomal anomaly, with a worldwide incidence of 1 per 1000-1100 live births.[2,6] There is the presence of mental retardation (MR) and multiple anomalies. DS presents with a multitude of medical problems.[2,6,11] Both patients had MR, but case 1 had no cardiac anomaly, whereas case 2 had a cardiac anomaly, which hindered our tendoachilles lengthening as there was an anesthetic risk and we had no cardiac backup. However,

the patient had already received the cardiac treatment and was on regular medications.

Around 20% of DS have orthopedic issues, including upper cervical spine instability, scoliosis, hip disorders like slipped capital femoral epiphysis, Legg-Calve-Perthes disease and patellar instability, and hand and foot deformities, and these are attributed to increased body mass index (BMI), muscular hypotonia, ligamentous laxity, and ankle instability.[1-3,7] Our cases had no scoliosis or hip problems.

Foot and ankle problems are commonly reported by DS patients but are still given less priority, though they have an effect on their gait.[3] Commonly reported foot deformities in DS are pesplanovalgus, hallux valgus, increased space between the first and second toes (sandal gap), syndactyly, clinodactyly, and metatarsus adductus.[1,3,5,7] Pesplanovalgus or flatfoot is commonly reported in DS, with incidence ranging from 60% to 91% in different studies.[1,3,6] Clubfoot in DS is less reported. The pathoanatomy of this disorder is different and still has occurrence in some. Both our cases had bilateral clubfeet. A large case series of Ponseti Users Group had 9 cases (13 feet) of Clubfoot with DS; among them 6 were males and 3 females.[12] One study reported 8 patients with 15 club feet in the year 1995.13 A retrospective study done over the period of 18 years reported a mere 13 patients of DS with club feet.[4]

Clubfoot in DS presents around 1-2 months of age, and the treatment is started therein, but our case 1 reported at 1 month of age, whereas case 2 had delayed presentation at the age of 18 months.[4,12-15] For case 2, treatment could not be started at an earlier age as there was a loss to follow-up. In comparison to idiopathic Clubfoot, these have lower pirani and dimeglio scores.[4,12,14] Our cases also had low pirani scores of 4 and 5 and dimeglio scores of 14 and 12, respectively.

Ponseti method achieves plantigrade foot in 95% of cases of idiopathic Clubfoot and is maintained in most of these cases, but the result has not been reported in Clubfoot in DS.[16] All the reported cases of Clubfoot in DS have undergone ponseti casting, and both cases also underwent the ponseti casting technique.[4,12-15] Both of our cases had supple Clubfoot and achieved the mid-foot score correction after 3 and 5 casts, respectively. Case 1 underwent TA tenotomy and correction, though we could not proceed with the same surgery for case 2, so we cannot comment on the outcome of that case. The number of casts used in different case reports/series is like ours.[4,12-15] So the ponseti cast achieves the plantigrade braceable foot in Clubfoot in DS. A study reports that Clubfoot with DS is not correctable by nonoperative treatment and will eventually require surgery for an acceptable result.[13] But some cases in this series had associated arthrogryposis, and stretching protocol might have also been different at that time, so casting alone could not achieve full correction. On the contrary, others have reported better results with ponseti casting alone.[4,5,8,9,12,14] Most cases required TA tenotomy. They had a similar rate of recurrences as compared to an idiopathic group but required less intraarticular surgery for its treatment.[4] Similar result was shown by a study in the English population.[12]

A further prospective study on a large scale is required to confirm the effectiveness of the ponseti method in Clubfoot associated with Down syndrome. A large study to find out the prevalence is also required.

CONCLUSION

Clubfoot may also be associated with Down syndrome, which is supple than the idiopathic counterpart and can be treated with the ponseti method and tendoachilles tenotomy.

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