

CTEV Deformity in Down Syndrome: A Rare Combination

Vinay Goyal¹, Sucheta Saha², Sandeep Kumar Gupt³, Nonica Laisram⁴

¹Associate Professor (PMR), All India Institute of Physical Medicine & Rehabilitation (AIIPMR), K.K. Marg, Mahalaxmi, Mumbai 400034.

²Consultant, Department of Neurorehabilitation, Institute of Neurosciences Kolkata

³Senior Resident, AIIMS Bhopal, Saket Nagar, M.P.

⁴Professor, Consultant & Head, Department of Physical Medicine & Rehabilitation, Vardhman Mahavir Medical College & Safdarjung Hospital, Ansari Nagar (East), New Delhi-110029.

Corresponding Author: Sucheta Saha

ABSTRACT

The incidence of Congenital Talipes Equino Varus (CTEV) is approximately one in every 1000 live births. CTEV has been associated with many neuromuscular and congenital conditions. Down syndrome is the most common autosomal chromosomal anomaly. A 2 year old boy with Down syndrome was referred to the department of Physical Medicine & Rehabilitation of a tertiary care Hospital, for bilateral feet deformity. On examination, we found feet deformities typical of CTEV. The co-existence of these two conditions is rare, as their basic pathologies are entirely different.

Keywords: Clubfoot, CTEV, Down syndrome.

INTRODUCTION

Down syndrome is the most common autosomal chromosomal anomaly. The orthopedic features usually seen are pes planus, atlanto-axial subluxation, patellar subluxation or dislocation, metatarsus varus, scoliosis, slipped capital femoral epiphysis and subluxated or dislocated hip joint. Patients with Down syndrome are characterized by- generalized ligament laxity. [1] We recently saw a patient with Down syndrome with CTEV in our department. In contrast to generalized laxity seen in Down syndrome resulting in pesplanus, CTEV deformity in Down syndrome is striking as basic pathology of clubfeet results in contracture of soft tissue, tendons and ligaments surrounding the

ankle, subtalar and midtarsal joints producing the deformity.

CASE REPORT

A 2 year old first order male child born out of Non-consanguineous marriage to a 24 year old father and a 20 year old mother with physical features of Down syndrome was referred to us for bilateral feet deformity. Antenatal period was uneventful. There is no family history suggestive of similar congenital deformities.

Clinical examination showed findings of Down syndrome, including-Mongoloid slant of eyes, epicanthic folds, depressed nasal bridge, protruding tongue, short neck, low set ears, stunted growth, delayed milestones, speech delay, mental retardation and hypotonia (Figure 1,2). The GTG chromosome banding analysis revealed 47XY with Trisomy of 21, diagnostic of Down syndrome. Patient was ambulant with manual support, could only turn 2-3 pages of the book at a time and came when called for, but indication for toileting needs has not started. He could only speak bisyllable words. His Developmental Quotient was 65. His echocardiography, Ultrasound whole abdomen and thyroid profile were normal.

Local examination of feet revealed typical appearance of clubfeet including-equinus of feet, adduction of forefeet and varus deformity of midfeet bilaterally with

the Pirani scoring of 3.5 on the right and 3 on the left side (Figure 3,4,5). No other orthopedic anomaly was detected in child. X-ray of whole spine was normal. Plain X-ray of both feet in Antero-Posterior and Lateral stressed dorsiflexion view- for measurement of angles of CTEV (Figure 6)

was done. The values are shown in the table:

Angles	Normal [2]	Right	Left
Talo-calcaneal (AP)	30-55°	18°	25°
Talo-calcaneal (Lateral)	25-50°	15°	15°
Talus-1 st metatarsal (AP)	5-15°	-8°	-10°
Tibio-calcaneal (Lateral)	10-40°	-35°	-33°



Figure 1



Figure 2

Patient showing typical features of Down syndrome: Mongoloid slant of eyes, epicanthic folds, depressed nasal bridge, protruding tongue, short neck, low set ears



Figure 3



Figure 4

Characteristic appearance of Clubfeet showing Equinus, Adductus, Varus & Cavus

Treatment for CTEV was started with manipulation and casting by Ponseti method. After two and a half months correction was achieved. Following correction patient was put on bracing. For delayed milestones- gait training in parallel bars, activities to improve hand functions and milestones, along with speech therapy were advised to the patient.



Figure 5: Unique combination of Down syndrome and CTEV



Figure 6: X-ray of both feet in Antero-Posterior and Lateral stressed dorsiflexion view- for measurement of angles of CTEV

DISCUSSION

Incidence of clubfoot is approximately 1 in every 1000 live births. Most cases are sporadic occurrences, although families with autosomal dominant trait with incomplete penetrance have been reported. [2] The basic pathology of CTEV is contracture of ligaments in the talonavicular joint complex.

Incidence of Down syndrome is 1/600-800 births. [3] The commonest orthopedic foot manifestation seen in Down syndrome is flexible pes planus as a result of ligament hyperlaxity. [4,5] As the basic pathology of these two conditions results in two different clinical pictures, their coexistence is rare. On review of literature we found a similar case series of 8 cases from USA [1] and a case report from Thailand. [6] Case report described a 1 month old girl of Down syndrome with bilateral CTEV. On chromosomal banding, they found translocation at long arm of chromosome 21 and the patient had cardiac abnormalities on echocardiography. We are presenting a similar case report of CTEV with Down syndrome in a 2 year old male child. But our case has trisomy of 21 and no cardiac abnormality.

Clubfeet is seen in Neurological conditions like Polio, Cerebral Palsy and Myelomeningocele and syndromes such as

Amniotic band syndrome, Arthrogryposis, Pierre Robin syndrome, Prune belly syndrome and many more with no mention of Down syndrome. [7]

CONCLUSION

In our daily practice of Rehabilitation Medicine, we encounter both Down syndrome & CTEV cases regularly but their coexistence is rare. CTEV if found in Down syndrome should be promptly managed for optimum rehabilitation of a child with Down syndrome. We already know that clubfeet is seen in various neurological conditions. Our experience from above case shows that it can also occur in Down syndrome.

REFERENCES

1. Miller PR, Kuo KN, Lubicky JP. Clubfoot deformity in Down's syndrome. *Orthopedic* 1995; 18: 449-452.
2. Beauty JH, Congenital Anomalies of the Lower Extremity. In: Campbell's Operative Orthopedics. Canale ST, Beauty JH, editors. Philadelphia, Pennsylvania: Mosby Elsevier, 2008:1063-1179.
3. Descartes M, Carroll AJ. Cytogenetics. In: Nelson Textbook of Paediatrics. Kliegman RM, Behrman RE, Jenson HB, Stanton BF, editors. Philadelphia: Elsevier, 2008: 502-516.
4. Diamon LS, Lynne D, Siman B. Orthopedic disorders in patient with Down's syndrome. *OrthopClin N Am* 1981; 12: 56-71.
5. Livingstone B, Hirst P. Orthopedic disorders in school children with Down's syndrome with special reference to the evidence of joint laxity. *ClinOrthop* 1986; 207: 74-76.
6. Mulprek P, Jirasirikul A. Down's syndrome presented with clubfoot deformity: A case report. *J Med Assoc Thai* 1999; 82(12): 1254-1256.
7. Janicki JA, Narayanan UG, Harvey B, Roy A, Ramseier LE, Wright JG. Treatment of Neuromuscular and Syndrome-Associated (Nonidiopathic) Clubfeet using the Ponseti Method. *J PediatrOrthop* 2009;29:393-397

How to cite this article: Goyal V, Saha S, Gupt SK et.al. CTEV deformity in Down syndrome: a rare combination. *Gal Int J Health Sci Res.* 2020; 5(2): 121-123.
