

Pediatric case of bilateral tibia varum with pseudoarthrosis & macrocephaly-primary report

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Abstract

This report describes a case study of a two and half years old male child with bilateral tibia varum and non-united fracture at the left tibia, referred for Physiotherapy. He was a term child, born to a NCM couple with no neurological complications. No history of early sepsis or systemic complications were reported. All milestones were attained in time. Child had age related social and communication skills. On evaluation, he has Macrocephaly, multiple cafe au lait spots, freckles at groin and axilla. This case was diagnosed as Neurofibromatosis Type 1. Physiotherapy objectives and plans are described and are open for discussion.

Keywords: tibia varum, psuedoarthrosis, neurofibromatosis

Introduction

Master Mohammed Riyan was referred to us from the Department of Physical Medicine and Rehabilitation, Govt District Hospital, Tirur, Kerala for Physiotherapy evaluation and intervention for his gait-related issues. We received the case on the first week of July 2021 in our outpatient unit.

Riyan is now two and half years old, very active and has age-appropriate communication skills. He was cooperating very well for Physiotherapeutic evaluations.

Brief History

He was born as the second child of a non-consanguineous marriage. No significant foetal complications were reported in the first two trimesters of pregnancy. It was during the third trimester, an ultrasonography picked up his bilateral bony deformities & above average head size.

He was born out of a caesarean section and cried immediately after birth. APGAR scores were good and reported birth weight was 3200 grams. On neonatal evaluation, doctors noted bilateral tibia varum and fracture of left tibia at the distal one third. No other congenital / systemic indications for early NICU interventions were noted.

Later, after detailed evaluation of the legs, the orthopedic doctors decided to go for conservative management, and opted serial casting upto one and half years. However, no remarkable progress were seen with casting. The fracture in the tibia remained unhealed (nonunion / pseudarthrosis).

Earlier this year, after a careful re-evaluation and board discussions by a team of specialists, Riyan is now advised to go for corrective surgical interventions of tibia bilaterally. He is now referred to the Physiotherapy unit for pre-surgical conditioning.

Observations and Findings

Riyan is a normally developing child, and has attained his gross, fine and social developmental milestones on time. These days he can walk with support using leg splints and walker rollator. He has an above-average head size (Macrocephaly). (Pic 1) No history of seizures or HMF dysfunctions are reported. No spine deformities are noted.



Fig 1: Macrocephaly

We noted generalised hypotonia, greater in the trunk and girdles. His lower limbs were internally rotated from hips. Bilateral hip abductors, external rotators, and extensors were weak. Remarkable bilateral tibia varum and non-united fracture of left tibia were seen. (Pic 2) And, as a result of pseudoarthrosis in the left tibia, his lower limb was apparently shortened by 3.4 centimetres.



Fig 2: Bilateral tibia varum and psuedoarthrosis of left tibia

On further evaluation, he is a wheatish brown complexion Indian boy with many dark coloured spots (cafe au lait spots) on the face, neck, upper limbs and trunk. Few freckles were noted in the groin and near axilla. (Pic 3) No subcutaneous nodes or bumps were felt.



Fig 3: cafe au lait spots

His vision and hearing functions are normal. No headache, tinnitus or related ear disturbances were reported by the parents. We could not perform any objective balance or proprioception scaling, however, based on our clinical experiences, we noted age appropriate balance skills in the child. Based on the clinical picture, Riyan has classical symptoms of Neurofibromatosis Type 1.

Neurofibromatosis:

These are a group of genetic disorders that cause tumors to form on nerve tissue (brain, spinal cord and nerves). Mutations of NF1 gene lead to the production of a non-functional version of neuro fibromin, that cannot regulate cell growth and division. As a result, tumors such as neurofibromas can form along nerves throughout the body later in life. There are three types of neurofibromatosis: neurofibromatosis 1 (NF1) diagnosed in childhood, neurofibromatosis 2 (NF2) diagnosed in early adulthood and schwannomatosis which usually affects people after age 20. The tumors associated with neurofibromatosis are often benign and slow growing. Apart from the clinical pictures, additional tests like ear test, balance test, imaging, genetic test are needed to diagnose NF. There isn't a cure for neurofibromatosis, but signs and symptoms can be managed medically or surgically.

Management Objectives and Plans

Based on the available medical reports, discussions, observations and findings, we prioritised and enlisted the following objectives and plans for master Riyan.

1. Since the child was suffering recurrent falls due to in-toeing associated with bilateral hip internal rotations, priority was given to address and reduce this by strengthening hip extensors, abductors and external rotators. (Pic 4) Both closed and open kinetic exercises were chosen for this.



Fig 4: Internal rotation of bilateral hips resulting in in-toeing

2. Second priority was given to hypotonia of trunk and girdles. Exercises on gym balls, upper limb weight shifts, exercises mimicking pelvic squaring, exercises in all four, crawl and reach, bridging, kneeling, prone lifts, were taught to the parents as home exercises program. They were advised to keep an exercise diary to monitor the performance.
3. Gait training using the leg splints needed corrections. Height of the walker rollators was increased above hip so that the child extends the hips and trunk better.
4. External reduction of the left tibial fracture and splinting was taught to the parents. This automatically adjusted the limb lengths.
5. General resistance exercises using resistive bands, weight jackets, cuffs, hydrotherapy (leisure activities in pool) were advised to improve general tone and strength. Activities on trampoline, balloon / bosu ball were avoided.

Since the parents were following the instructions well, the child is now kept on a home based exercises program with regular medical and Physiotherapy reviews. Moreover, we monitor the home based exercises regime through video calls. Apart from these interventions, the parents were advised to watch for secondary complications like scoliosis, hearing or visual deficits, neurological regressions etc. Our hopes in this child stand strong and we believe, in the next four months, Riyan will be fit for the corrective surgeries. Accordingly, his progress will be reported.

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