Scholars Journal of Medical Case Reports **ISSN 2347-6559 (Online)**
Sch J Med Case Rep 2015; 3(7):574-576 **ISSN 2347-9507 (Print)**

(An International Publisher for Academic and Scientific Resources)

Physiotherapy in Caffey's disease: a Case Report

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Abstract: Caffey's disease also known as Infantile Cortical Hyperostosis is rare and self limiting condition affecting young infants causing acute inflammation of periostium, soft-tissue swelling, and irritability. As inflammation subsides, certain musculoskeletal impairments take over. Reduced joint range of motion due to muscle tightness, if not treated on time can lead to contractures and long term impairments. As infants suffer from this disease, because of their musculoskeletal impairments there is a difficulty in their functional activities which lead to delay in achieving age appropriate motor milestones. This can be successfully managed by early diagnosis and prompt physiotherapy treatment. The present case report emphasizes on the musculoskeletal physiotherapy treatment in Caffey's disease in three months old infant. Caffey's disease being one rare disease, emphasis here is to increase awareness regarding the disease entity, its musculoskeletal impairments leading to functional issues and physiotherapy management.
Keywords: Caffey's disease, infantile cortical hyperostosis, Physiotherapy.

INTRODUCTION

Caffey's disease is a rare condition originally described by Caffey and Silverman in 1945[1]. It is also known as infantile cortical hyperostosis, is a self-limiting disorder that affects infants and causes bony changes, soft-tissue swelling, and irritability [2]. The diagnosis is commonly delayed as this disorder mimics a wide range of diseases including osteomyelitis, hypervitaminosis A, scurvy, bone tumors and child abuse [3]. As inflammation subsides, certain musculoskeletal impairments like muscle tightness, reduced range of motion may take over. The present case report of Caffey's disease emphasizes on physiotherapy treatment to improve mobility, to achieve age appropriate motor milestones, and facilitate functional development. Aim of this paper is to increase awareness regarding the disease entity, its musculoskeletal impairments leading to functional development and physiotherapy management. Prolonged immobility leads to muscle tightness, which in long term may cause contractures and thus reduce joint range of motion. These impairments hamper motor development of a child affecting functionality. Proper Physiotherapy management in terms of stretching, myofascial release, joint mobility techniques and functional training will facilitate development of a child by emphasizing on a complete patient care.

CASE REPORT

History and Examination

Three months old Indian male child of non-consanguineous parents, born full term with normal delivery and was exclusively on breastfeeding presented with suddenly developed irritability with mild fever. Mother noticed reduced movement of left lower limb compared to right side and swelling over left thigh region. There was no history of trauma or positive family history of similar condition. Clinical examination showed uniform swelling of entire left thigh which was non-tender, firm in consistency. Laboratory investigations showed Elevated Erythrocyte sedimentation rate and alkaline phosphates levels which were 95 mm/hr and 254 U/L respectively. Radiological findings revealed femoral periosteal hyperostosis with cortical thickening. Open biopsy showed hypercellular reactive new bone formation in femur and absent malignant cells. This ruled out malignancy. Pediatric orthopedic surgeon made the diagnosis of Caffey's disease on the basis of the presentation and investigations. After starting with medical management with NSAID, baby was referred to physiotherapy at the end of three months.

On Physiotherapy assessment an adherent surgical scar of the biopsy was present on anteromedial aspect of upper thigh. The iliopsoas and hamstrings were severely tight along with affected range of motion in left lower limb as follows:

Table-1: Range of motion in left lower limb

Hip flexion	40º - 70º
Hip Abduction	0- 20º
Knee flexion	20º - 140º

Functionally as per his age of 7 months expected milestones were rolling on both sides, supported sitting, scoot, rock back and crawling. When he was referred for physiotherapy, he had delay in these milestones due to lower limb involvement. Rolling from side-lying to prone, crawling and sitting was not achieved due to joint mobility limitation.

Intervention

Treatment started with scar mobilization including kneading and friction massage, stretching with myofascial release for hamstrings and iliopsoas to improve joint ranges. Strengthening exercises in terms of kicking a toy, holding a ball between thighs and legs were given. Special emphasis was given for achieving motor milestones i.e. rolling and going to prone position followed by going on all four for crawling. Rolling was achieved with reflex rolling, prone to supine rolling with pelvic rotations on mat and on vestibular ball. Crawling was achieved with encouraging child to rise on knees, weight bearing on knees, forearms and knees. This was done with pressing the child's lower back and increase weight bearing on knees.

Treatment Outcome

Child started showing improvement in activities as range of motion improved. Supported sitting was achieved as the hip ranges improved gradually till 100º. Full hip and knee ranges and mobile scar were achieved at the end of one month of regular physiotherapy. Baby achieved all age –appropriate milestone and could crawl on all fours, come to sitting with support. On follow up after three months baby was playful and active with full ranges and had achieved unsupported sitting and could come to supported standing. There was no limb length discrepancy. Thigh swelling was also completely resolved. At six months follow up, baby was walking. There was no recurrence.

DISCUSSION

Caffey's disease is characterized by triad of soft tissue swelling, cortical thickening of the underlying bone and hyperirritability in narrow age group i.e. between birth and five months. The condition is being described as being rare with no sex or racial predilection [4]. The cause of Caffey's disease is not clearly known. Reports of familial occurrence suggest a possible hereditary defect of the arterioles of the periosteum [5]. Recent genetic research suggests COL1A1 gene mutation affecting collagen formation [6, 7, 8]. The swelling occurs suddenly, is deep, firm, and may be tender. In Caffey's disease, several parts of the body can get affected. Available data showed ulna was most commonly affected followed by mandible, tibia, clavicle, scapula and ribs[9].The humerus, femur, fibula as being less commonly affected; there is a delay in diagnosis and thus further management. Caffey's disease in this case was diagnosed on the basis of age i.e. three months, triad of irritability, swelling and excessive periosteal new bone formation seen on radiograph of femur. There was also insidious onset of swelling with no other signs of inflammation like local tenderness or redness which rules out infection. As baby was exclusively breast-fed, Hypervitaminosis A was excluded. Also Scurvy and Hypervitaminosis A occurs in infants above one year of age. Trauma and fracture was excluded as there was an excessive callus formation with no evidence of fracture in radiograph and no history of trauma. Tumors have onset of symptoms in weeks rather than days as in this case, but it was ruled out by open biopsy of femur.

The disease affects diaphysis of the bone and thus joint surfaces are spared [10]. However because of the immobility to reduce pain during the episode of inflammation, there is a reduction in joint mobility. Functionally, there is an effect on musculoskeletal system like muscle tightness and decreased range of motion. For relieving tightness, gentle sustained stretching, myofascial release are very useful. Due to musculoskeletal impairment, patient had difficulty in achieving motor milestones such as rolling and crawling which involved coordinated movements of hip-knee flexion and extension. Patient also had difficulty to come to sitting due to altered hip – knee ranges. As joint mobility improved with regular physiotherapy, child successfully improved. In conclusion, though Caffey's disease is a self-limiting disease; there can be certain musculoskeletal impairments hampering patient functionally. This case demonstrates good improvement in a case of Caffey's disease with physiotherapy. It provides clinical information for the therapist regarding existence of Caffey's disease and its physiotherapy implications.

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