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## Case Report

# Experience in Managing Child with Congenital Fiber Type Disproportion with Rehabilitation Outcome: Case Report

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**Background:** Congenital fiber type disproportion (CFTD) is a histopathological diagnosis, with non-progressive condition and varied clinical presentations. CFTD may cause significant disability due to muscle weakness and related comorbidities.

**Case report:** A 10-year-old boy with clinical presentations of general dystonia since birth was diagnosed as CFTD. His motor domains delayed but the neuropsychological aspects were within normal range. He underwent long term physical therapy and speech therapy since birth. The nasogastric tube was removed until he was 3 years old. The muscle strength and endurance improved along with age and after rehabilitation. Comorbidities include scoliosis, asymmetry of chest wall, musculoskeletal pain were managed. The activities of daily livings were partially involved in motor scale. Modification of life style and the concept of energy conservation were introduced.

**Conclusion:** Multidisciplinary interventions and health education help in managing the motor delay and functional regain. The goal may be individualized after comprehensive evaluation. ( Tw J Phys Med Rehabil 2018; 46(2): 101 - 106 )

**Key Words:** Congenital myopathy, congenital fiber type disproportion, case report

## INTRODUCTION

Congenital fiber-type disproportion (CFTD) is a rare, genetic, nonprogressive muscle disease that is diagnosed histopathologically and is a type of congenital myopathy. The general consensus used in diagnosis through muscle biopsy is the presence of at least 12% smaller type 1 muscle fibers than type 2.<sup>[1]</sup> CFTD principally manifests as muscle weakness that is generalized or limited to

proximal muscles. Comorbidities include face deformity, ophthalmoparesis, high-vaulted palate, muscle contractures, scoliosis, and even cardiac dysfunction<sup>[2]</sup>. CFTD can have similar pathological findings to other myopathies, and clinical presentation of CFTD is varied; making an accurate diagnosis is therefore both challenging and essential.<sup>[3]</sup>

This report presents a case history of a patient with CFTD undergoing long-term continuous rehabilitation during the first decade after birth.

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## CASE PRESENTATION

A 10-year-old boy born at full term presented with generalized hypotonia at birth, without structural abnormalities. Persistent difficulty during sucking was observed at first month after birth, which presented as frequent choking, and his parents thus sought pediatric evaluation. Neurological examination revealed no major abnormalities. His level of creatine kinase was 230 U/L, which was slightly higher than normal limit. Difficulty in chewing and swallowing were observed. A nasogastric feeding tube was inserted at 4 months as he was failing to thrive, probably in relation to poor feeding, and the tube remained until he was 3 years old. Recurrent episodes of pneumonia were recorded in the first 3 years, possibly related to the aspiration of secretions, but these did not necessitate tracheal intubation. At the age of 3 years, a muscle biopsy through the right rectus femoris muscle was performed, and histopathological findings supported a diagnosis of CFTD. However, he did not receive gene study for further survey. There was no family history of neuromuscular diseases.

He was referred for developmental evaluation because of his slow responses and poor motor function, and a diagnosis of developmental delay was confirmed at 8 months of age. At that time, his gross motor function has been found to be delayed, compatible with age of 3 to 5 months by using Early Intervention Developmental Profile. He began to walk when he was 1 year and 8 months old. When applying the Peabody Developmental Motor Scales–2nd Edition, we determined persistently delayed gross motor function and borderline delayed fine motor function. The Gross Motor Quotient was 70 at 3 year and 8 months old (borderline delayed), 59 at 4 years and 10 months old (<1 %, delayed). At age 6, a Chinese Version of Pediatric Evaluation of Disability Inventory showed marked decreased standard score, less than 10 (mean of 50, standard deviation of 10), indicated delayed in both functional skill and caregiver assistance aspects of self-care domain.

A Bruininks-Oseretsky Test of Motor Proficiency II was applied for evaluation of his fine motor function. At 4 years and 10 month, his fine manual control was at 50 % (within normal limit) , and the manual coordination was at 37%(well-below the average). While the fine manual

control was 92 % ( higher than average) and the manual coordination was less than 14% (well-below the average) at 6 years and 7 months old.

The IQ test and psychosocial evaluation were all found to be within the normal range at a serial follow-up. An intelligence evaluation presented as a full-scale intelligence quotient/percentile rank was conducted using the Wechsler Preschool and Primary Scale of Intelligence–Revised (WPPSI-R), and the results were 106/66 at 3 years and 8 months, 105/63 at 4 years and 10 months, and 117/87 at 6 years and 7 months. The most recent IQ test was taken after attending school when he was 9 years old, and the result was 89/32. A progressive lower performance scale was observed in the visual-motor coordination and processing speed.

Regarding the speech evaluation, his comprehension was within normal limit, however, disarticulation was observed. The presentations were hypernasality, omission, and aspiration of substitution. The presentations may be the consequence of oropharyngeal muscles weakness.

Based on the evaluation finding, he underwent physical therapy, occupational therapy, and speech therapy in early childhood, prior to confirmation of CFTD diagnosis. Speech therapy was applied for oral motor function, particularly in relation to articulation and swallowing function.

The patient showed typical facial features of myopathic subjects, with an increased lower facial height and high arched palate. He regularly attended dental outpatient clinics for survey of dental carries and follow up development of dentition, while cross bite was observed in the serial follow up. Thoraco-lumbar scoliosis with asymmetric right pectus carinatum was observed at 3 years of age, and GYROKINESIS® was applied to correct his spinal and chest wall deformities. The initial Cobb's angle was 17°, but this decreased to lower than 10° after physical therapy. The profound asymmetry of his chest wall was also corrected.

Due to complaint of muscle pain and fatigue after ambulation in school, we examined the daily activities and discussed the energy conservation strategies with him and his parent. He was advised to use a wheelchair when moving over long distances during the daytime. Moreover, he was advised to perform normal daily activities when possible at home, but such activities must be prioritized. A

light-weight foot orthosis was applied to his profoundly pronated foot to increase his ambulation ability, and improved stability and decreased postural sway were observed. A school entry delay of 1 year was suggested after evaluation, in consideration of his physical condition and the accessibility of a suitable educational environment.

He complained of joint pain after vigorous and/or prolonged activity. His muscle weakness and related imbalance also caused occasional fall injuries, which subsequently led to multiple joints and musculoskeletal pain. Although the pain usually resolved after conservative treatment, when he was 7 years old, he knocked his sacral region in a fall during transit, and the associated pain persisted for nearly 6 months without any radiographic evidence of bony injury. This pain hindered his daily activities and the rehabilitation program. Although he regained physical activity after a prolonged rest, his body weight significantly increased during the period of immobility. He is currently able to climb two floors of stairs or walk 50 m continuously; however, muscle pain and knee joint pain have become increasingly severe when climbing stairs.

The patient continues to have good peer relationships and his academic performance is above average at school. He is currently partially assisted with activities of daily living (ADL). He scored 104 (out of a total score of 126) in the WeeFIM test, which was used to assess his ADL. Subscale results were 71 of 91 on the motor scale and 33 out of 35 on the cognitive scale. Of all domains, his self-care (including bathing and dressing) is most affected. The Child Health Questionnaire (CHQ)<sup>TM</sup> parent form (CHQ-PF50) was applied to evaluate his physical and psychosocial conditions<sup>[4]</sup>, and his parents reported responses. The score comprises 12 domains and is transformed into two main scores: a summary score (PhS) and a psychosocial summary score (PsS). The patient scored 14.92 in PhS and 56.52 in PsS (mean of 50 and standard deviation of 10). Both results in WeeFIM and CHQ-PF50 suggested that his physical performance was more affected than psychosocial aspect, reflecting the essence CFTD affected physical function predominantly.

## DISCUSSION

Despite the debate existed whether CFTD was a di-

agnosis or syndrome<sup>[5]</sup>, it was still regarded as a diagnosis that persistently used in clinical practice. CFTD is a rare but nonprogressive disease<sup>[6]</sup>. The rehabilitation goal for pediatric patients with CFTD is to attain normal milestones through multidisciplinary intervention. The concept of energy conservation is important in the treatment of myopathic patients. Due to an imbalance in energy expenditure, training highly depends on an adequately distributed program focusing on daily life, with a decreased focus on non-functional or non-goal training<sup>[7]</sup>. Generally, rhythmic and aerobic exercise, such as cycling, hydrotherapy, or swimming are suggested in patients with congenital myopathy<sup>[3]</sup>. However, while the patient was undergoing treatment for chest wall asymmetry and scoliosis, rehabilitation focused mainly on specific exercises instead of endurance or aerobic training because the patient got tired easily. The patient and his parents were provided with ample health education, and were able to organize his schedule according to his expected energy expenditure and prioritizing events.

A 6-min walking test is conducted when evaluating the physical fitness of pediatric patients with myopathy<sup>[8]</sup>. Other evaluation programs are also often proposed, such as a gait analysis or the Timed Up to Go Test<sup>[9]</sup>. However, such tests were not conducted with this patient because of his poor endurance and lack of confidence in undertaking physical tests. Observations of his progression were undertaken and intermeasurement differences of fitness (such as the duration and distance achieved when pedaling or walking) were conducted to evaluate cardiopulmonary fitness. Our patient has shown limited progressive improvement in his endurance ability throughout the period of observation, and in the most recent year he has been recorded as being able to pedal 400 m and walk 50 m within a certain time frame. Although he still presents with poor endurance, further exercise programs have been administered to maintain endurance and body fitness.

Dysphagia is a common problem in patients with CFTD<sup>[10]</sup>, due to involvement of swallowing muscles, and an early tube-feeding or gastrostomy are often required in accordance with the poor functioning of such muscles. A nutrition consultation was provided to the parents of the patient, and rehabilitation intervention focused on dysphagia, oro-motor exercises, and phonetic placement; positive responses were observed to these

early interventions. As mentioned, he initially suffered from severe dysphagia and was fed through a nasogastric tube from the age of 4 months until 3 years (when he began oral feeding). Dysphagia not only affected the nutrition, but also caused repeated pneumonia, possibly related to aspiration of food or secretions. After observation his improvement of swallowing function and resolution of pneumonia, the nasogastric tube had been successfully weaned. Currently, his body weight is within the normal growth range and he tolerates soft food through oral feeding; however, he cannot manage crude fiber or solid food.

Myopathy-related scoliosis is another presenting issue for these patients. In his case, mild kyphoscoliosis and profound right pectus carinatum were noted at 3 years of age. Physical therapy focused on correcting the alignment of his spine and chest wall deformity. There is currently no common consensus on the best type of physical therapy to use with children who have neuromuscular scoliosis. However, in our case, intervention with GYROKINESIS® was decided with the patient's agreement. In serial follow-ups, the angle of scoliosis was found to have decreased and the asymmetry of the bilateral chest wall had significantly improved, indicating the advantage of early intervention with respect to plasticity.

A further problem facing these patients is the presence of myalgia or muscle pain. Most of our patient's pain episodes were related to prolonged muscle use or overuse<sup>[9]</sup>. Conservative treatment strategies such as applying an ice pack, resting, or taking local NSAID medication have been effective. However, it is necessary to remember that muscle pain in a myopathic patient is not always related to muscles but can be related to orthopedic or rheumatological conditions<sup>[11]</sup>. In our case, we considered possible orthopedic diseases to explain his clinical condition after one episode of sustained pain. Patellofemoral syndrome was diagnosed after comprehensive physical examinations and imaging. An insole was provided to correct an overt pronated foot by adjusting his biomechanical misalignment. Besides, the patient presented with joint hypermobility, which suggested that altered joint dynamics and decreased stability that susceptible to the musculoskeletal injury. Though hypermobility was not uncommon in patient with congenital myopathy<sup>[12]</sup>, the phenomenon of hypermobility was not

yet mentioned in CFTD before. Moreover, joint dislocation is an issue that could occur in certain congenital myopathy<sup>[13]</sup>, however, there was no literature that reported the increased tendency of joint dislocation in patient with CFTD. This occurrence shows that any prolonged subjective complaint of pain should not be neglected, and comprehensive evaluation should be performed.

Children with CFTD present with different disease severity levels. More severe types can involve profound respiratory muscle weakness, involve facial and bulbar muscle weakness, and necessitate the use of a feeding tube or gastrostomy. Such symptoms are considered predictors of a poor outcome<sup>[14]</sup>. However, Tsuji et al. described the marked improvement of a patient with a severe form of CFTD, which again suggests the potential of a patient reaching near-normal performance<sup>[15]</sup>. Our patient, who has a mild-to-moderate form of CFTD, showed improvements in his functional achievements, which reflects the nonprogressive characteristic of congenital myopathy. Multidisciplinary interventions and health education assist in managing motor delay and functional regain. However, several symptoms, such as neuromuscular scoliosis and/or dentalfacial problem, can gradually present, and conservative treatment is the main initial treatment choice. In addition, further invasive procedures, such as orthognathic surgery or scoliosis surgery, may be required depending on the individual's functional status and motivation.

## CONCLUSION

This case report presents the rehabilitation and long-term follow-up of a child with CFTD. Patient with CFTD was affected mostly in the physical function and subsequently decreased ADL activities. Rehabilitation was mainly in managing the motor delay and functional regain.

Monitoring for further development of musculoskeletal injuries, neuromuscular scoliosis, and dentalfacial issue is warranted. It is important that patients with CFTD attain individualized goals, and a combination of comprehensive evaluation and care are suggested.

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## DECLARATION OF INTEREST STATEMENT

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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# 處理先天性肌纖維類型不均衡病童之復健經驗：個案報告

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Congenital fiber type disproportion(CFTD)為一非進行性先天性肌肉病變，須經肌肉切片病理確診，其特點為第一型骨骼肌纖維體積比第二型肌纖維至少小 12%。現有文獻提及此疾病之臨床表現變異性大，除肌肉無力，可能併有弓狀顎 (High-arched palate)、眼肌麻痺症 (Ophthalmoplegia)、呼吸障礙等。患者皆因肌肉無力造成顯著功能障礙。本案例為一 10 歲確診 CFTD 男童之長期追蹤與復健介入之描述。

患者出生時明顯肌張力不全，自四個月起因生長遲滯、吞嚥功能障礙而接受鼻胃管置入，但診斷未明；後續持續安排物理治療與語言及吞嚥治療，該患者於一歲八個月時開始走路，三歲時成功移除鼻胃管，並於此時才確診 CFTD。

患者定期接受完整發展評估，結果顯示明顯動作發展遲緩，其社會心理領域評估結果皆為正常範圍。該患者的肌力與肌耐力隨著年紀增大與物理治療介入而有進步。日常生活能力評估於運動部分仍部分依賴。

此外，長期追蹤過程發現該患者有多種肌肉病變之併發症，包括脊柱側彎、雞胸(pectus carinatum)、以及關節與肌肉疼痛。物理治療介入以及教導患者以及家屬運用節省能量之日常生活方式，包括輪椅使用、自我安排活動。

本病例報告紀錄 CFTD 肌病變患者之長期追蹤與復健過程，根據患者之狀況設定個人化復健目標，及處理相關之併發症。(台灣復健醫誌 2018；46(2)：101 - 106)

**關鍵詞：**先天性肌肉病變(Congenital myopathy)、先天性肌纖維類型不均衡(Congenital fiber type disproportion)、病例報告(case report)