

Whistling-face Syndrome – A Case Report

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Abstract

The craniocarpotarsal, or “whistling face” syndrome was first described by Freeman and Sheldon in 1938. It’s an extremely rare condition, comes under one type of distal arthrogryposis category. Prominent deformities include deformity of hand and foot with typical whistling face. Early diagnosis of the condition aware the clinician about resistance to different therapeutic manoeuvres and management is planned accordingly.

Key words: Arthrogryposis, Freeman–Sheldon syndrome, craniocarpotarsal dystrophy.

Introduction:

Whistling-face syndrome (WFS), is characterised by craniocarpotarsal dystrophy. It was originally described by Freeman and Sheldon in 1938¹, also called as Freeman–Sheldon syndrome. It is a rare form of multiple congenital contracture (MCC) syndromes (arthrogryposes multiplex congenita) and is the most severe form of distal arthrogryposis (DA)^{1,2}.

As per presentation of the condition, different terminologies assigned for the condition are distal arthrogryposis type 2A (DA2A), craniocarpotarsal dysplasia (or dystrophy), craniocarpotarsal syndrome, Windmill-Vane-Hand syndrome.

Distal arthrogryposis was identified as a separate genetic disorder in 1982³. Characteristically distal part of the limbs i.e. hands and feet are involved. Sometimes

proximal affection like congenital knee flexion contracture and hip dysplasia are seen. There are three forms of DA; DA1, DA2A, DA2B. DA1 is the least severe; DA2B is more severe with additional features that respond less favourably to therapy. DA2A (WFS) is the most severe of the three, with more abnormalities and greater resistance to therapy².

The diagnostic criteria for DA2A or WFS includes two or more features of DA: microstomia, whistling-face, nasolabial creases, and ‘H-shaped’ chin dimple¹ (Fig 1). The condition is also described as a type of congenital myopathy⁴.

Besides the craniofacial manifestations patients generally seeks for deformities of hands and feet. Common hand deformities are clenched hand or thumb in palm deformity and foot present with resistant club foot deformity. Virtually all individuals with DA are born with their hands clenched tightly in a fist due to the abnormal muscle physiology⁴. The deformities are very much resistant to therapeutic stretching and serial corrective plaster cast. Difficulty in endotracheal intubation and predisposition to malignant hyperthermia and frequent respiratory tract infection in these cases also increase anaesthetic risk for surgery. Unfortunately, many surgical procedures have suboptimal outcomes, secondary to the myopathy of the syndrome.

Case Report:

A 6 years boy admitted to Department of Physical Medicine and Rehabilitation for difficulty in walking and difficulty in holding objects because of deformity of hand and foot from birth. None of his family member

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Fig 1- Whistling-face Syndrome with H-shape Chin Dimple

had any form of congenital limb anomalies. There was no pre or perinatal bad history. There was no known history of maternal exposure to drug or radiation. The mother and child were immunised properly as per WHO guidelines.

The child was thin built, short stature, small orifice of mouth (microstomia), nasolabial creases, prominent supraorbital ridges, typical chin dimple and whistling-face (Fig 2). Speech was not cleared. Both the hands



Fig 2- The Same Child with Hand Deformity

presented with similar deformity with thumb adducted and flexed over palm and other fingers overlapped over thumb like a clenched fist. There was no voluntary opening of fingers. Both the feet were operated for club foot deformity 2 years back and recurrence of deformity on both the sides. Deformity on left side was much rigid (Fig 3). Abduction of on left side was limited. Both the lower limbs were cylindrical shaped with less subcutaneous fat as seen in arthrogryposis. The child was dependent for most of his ADL.

All the blood parameters were within normal limit. X-ray of skull and spine did not show any specific abnormality. Left side dysplasia of hip was marked in x-ray of pelvis. After anaesthetic clearance the child had undergone surgery for correction of his rigid left club foot deformity by universal mini external stabilisation system (UMEX) (Fig 4). Hand deformities were tried to manage conservatively by stretching and adaptive devices.

Discussion:

WFS is an uncommon congenital anomaly mostly found in journals of genetic studies. There is paucity of clinical literature on WFS.

So far 65 patients have been reported in the literature, with no sex or ethnic preference notable^{2,4}.

WFS is caused by genetic changes. Krakowiak *et al.*^{5,6} mapped the DA multiplex congenita (DA2B; MIM #601680) gene, a syndrome very similar in phenotypic expression to classic WFS, to 11p1 5.5-pter. Other mutations have been found as well^{7,8}.

Toydemir *et al*⁹ showed that mutations in embryonic myosin heavy chain 3. In 1996, more strict criteria for the diagnosis of WFS were drawn up, assigning the syndrome as DA type 2A².

Due to the abnormal muscle physiology in WFS, therapeutic measures may have unfavourable outcomes⁴, deformities recurred very often even after adequate correction¹⁰. Patients and their parents must receive psychotherapy, which should include marriage counselling¹¹. Chronic psychological problems, including depression secondary to chronic illness of unfavourable outcomes, can be very successfully addressed with early interventions¹². The child should have pre-emptive and ongoing mixed cognitive therapy- psychodynamic psychotherapy for patients with WFS and cognitive-behavioural therapy (CBT), if begun after onset of obvious pathology.



Fig 3- Deformed Left Foot

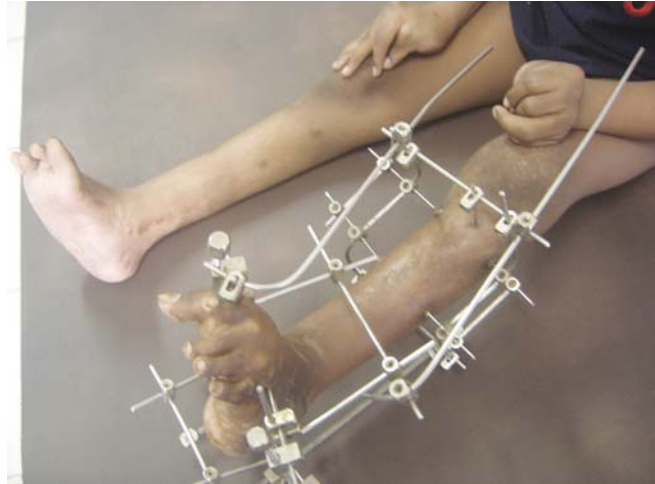


Fig 4- Corrected Left Limb

A family with 7 persons affected with WFS in 3 successive generations has been described by Wettstein and Buchinger¹³. Six affected persons are female; the only male carrier died in early infancy because of the severity of symptoms.

All efforts to be made to make the child ambulatory by correcting the foot deformities.

There are little data on prognosis. However respiratory complications are very common even death from pneumonia, empyema have been reported^{14,15}.

Conclusion:

WFS is an uncommon congenital anomaly. Awareness of such a resistant syndrome is useful in clinical practice of a physiatrist. Since prominent manifestations being the deformity of hands and feet, making the child dependent on ADL, early setting of rehabilitation goal is essential. The objectives of management would be directed towards ambulation of the child and improvement of hand activities. Early intervention gives better result with special emphasis on chest therapy. Psychotherapy for patient and parent is an integral part of rehabilitation.

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