



Podiatric Features of Velocardiofacial Syndrome: A preliminary study

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Abstract

In this study, 12 patients with a diagnosis of Velocardiofacial Syndrome and a complaint of leg pains were clinically assessed for biomechanical lower limb abnormalities at Northampton School of Podiatry, Nene College, Northampton, UK. The details of the history, the variety and prevalence of the diagnosed biomechanical abnormalities and the effect of podiatric treatment on the patients symptoms are reported.

This work suggests leg pains as a new feature of Velocardiofacial Syndrome and recommends podiatric measures as a non invasive method of treatment that achieves, at least, partial relief.

Background

Velocardiofacial syndrome (VCFS), also known as Shprintzen syndrome¹, is an autosomal dominant disorder², that results from deletions and microdeletions on the long arm of chromosome 22q,4,5. Deletions on this area are denoted by the phrase 22q11 where, 22 is the number of the affected chromosome, q refers to the long arm of a chromosome and 11 specifies the particular locus on which the deletion has occurred on the particular arm of the chromosome concerned.

Deletions and microdeletions on 22q11 may result in a number of clinical syndromes that bear certain similarities to one another. Examples of such syndromes include VCFS1, DiGeorge syndrome⁶, Conotruncal face anomaly syndrome⁷, Familial congenital heart disease⁸ and others. Among over 180 recognised clinical features⁹, the most common of VCFS include, cleft palate, congenital heart disease^{10,11}, characteristic facial appearance (Facial Dysmorphism)¹², thymus and parathyroid gland abnormalities¹³, learning and social difficulties¹⁴, and psychiatric disorder.^{15,16}

In June 1995, a 5 year old female VCFS patient was seen in a National Health Service Podiatric Clinic at Milton Keynes, UK, for the treatment of traumatic blistering of the dorsum of the lesser toes. The patient's mother complained that the child was suffering from recurrent episodes of leg pains. General Practitioners and Paediatricians found no apparent reason for such episodes. Sometimes a diagnosis of Growing pains was made and in other instances the parents were told that the child was not suffering from leg pains. Further inquiries revealed that this parent knew of at least another 4 VCFS patients who suffered similar episodes of leg pains. A thorough litera-

ture search revealed no information about leg pains in VCFS. By April 1997, 12 VCFS patients with leg pains were biomechanically assessed and podiatrically treated at the Northampton School of Podiatry Clinic, Northampton, UK.

History

The detailed history obtained from patients and parents revealed a similar pattern of pain complaint and pain behaviour suggestive of the presence of leg pains.

Only a proportion of the patients complained of leg pains (*see discussion*). On the other hand, pain behaviour (*explained below*) was observed in all patients of this sample. The pain was described by those who complained, as a burning or cramping sensation in the back of the legs. The severity of the pain seems to be perceived by most patients as moderate and has no specific radiation. It occurred mostly at night particularly at the end of a physically active day. The frequency of these episodes varied widely between patients from once or twice per week to over 9 episodes per day.

All parents reported two observations, which will be referred to as Pain Behaviour. A decrease in the tolerance to physical exercise was the first pain behaviour observed. This was manifested by the child needing a rest during a relatively short walk. The child may lag behind, demand to be picked up or even cry until picked up. Curiously however, some children were able to persevere with activities they prefer, like dancing, more than mundane activities like walking. This was confirmed during gait analysis when some parents expressed surprise at the distance the child was able to walk on the treadmill without needing a rest.

The second pain behaviour observation was the frequent episodes of waking up in the night. Again, only some of the children woke up complaining of pain and the frequency of these episodes varied widely between patients. In the majority of cases however, nocturnal episodes of leg pains proved troublesome. Children may wake up rubbing their legs, kicking their legs in a restless state, crying or sometimes wake up for no obvious reason.

Examination

A biomechanical examination of the lower limbs included 12 VCFS patients, 6 males and 6 females aged between 4 and 14 years in April 1997. The aim of the examination was to clinically survey the lower limbs for any biomechanical abnormalities that could explain the pain. All



Velocardiofacial Syndrome

examinations were carried out at the Northampton School of Podiatry Clinic by the same clinician, sometimes in conjunction with other clinicians. The most striking feature was that every patients in this sample showed one or more biomechanical abnormality of the lower limb. The table below shows the most common biomechanical deformities diagnosed and the frequency of their occurrence in this sample. (*Sample size = 12 patients.*)

Number of VCFS patients with each lower limb abnormality:

Type of deformity:	Number of patients:	Percentage:
Excessive STJ pronation	9	75%
Soft Tissue Ankle equinus	8	66.7%
Toe deformities	8	66.7%
Hypotonia	7	58.3%
Ant.disp.tib.post.tendon	6	50%

STJ = Subtalar joint.

Ant.Disp.Tib.Post.Tendon = Anterior displacement of tibialis posterior tendon.

Treatment

The aims of the treatment were to provide extra cushioning, prevent forefoot hypermobility, prevent abnormal compensation at the subtalar joint and to gradually stretch the calf muscles when applicable. The long term effect is to maintain a proper skeletal alignment and allow the individual to function much more efficiently.

All patients were treated with a simple insole made of a 3 mm poron (Open cell polyurethane foam) base with a poron pad. The type and the thickness of the prescribed pad depended on the type and the severity of the deformity. Patients with excessive subtalar joint pronation, were treated with a Cobra Pad (Rose Schwartz meniscus extended into a valgus rest)¹⁷ (Fig 1). Patients who showed soft tissue ankle equinus, were treated with a Heel Raise and muscle stretching exercises. Footwear advice was given to all patients and parents.

Response to treatment

Attempts to quantify the response to treatment through parents recording pre and post-treatment observations have unfortunately failed. Non of the parents recorded any observations. Nevertheless, through unstructured telephone interviews, all parents reported what they described as: 'A very noticeable improvement with a definite improvement in exercise tolerance and a decrease in the number of episodes of nocturnal leg pains.' Children who complained of pain, reported an improvement in the severity and frequency of pain episodes following the treatment.

Discussion

Some of the patients did not complain of leg pains, yet they showed pain behaviour suggestive of leg pains including exercise intolerance and nocturnal episodes. Furthermore, these features improved in response to treatment. This supports the suggestion that such patient are suffering from leg pains in spite of the absence of verbal complaint. The lack of a verbal complaint may be due to a communicative disorder, which is one of the recognised, not uncommon features of VCFS¹⁸. The patients' young

age may also be a factor. A number of parents suggested the possibility that the pain might have always been present, and therefore the child was not aware of any problems out of the ordinary hence, the lack of verbal complaint. Some parents were surprised to find that their non complaining child was suffering from leg pains. This finding clarified some of the childrens unexplained behaviours.

The early detection and treatment of pedal abnormalities in congenital chromosomal syndromes has been strongly recommended e.g. Downs syndrome¹⁹. In spite of the fact that leg pains might not be as serious as some of the other features of VCFS, yet it exerts an unfavourable effect on the quality of life of the patient and the family. Exercise intolerance adds to the social isolation of the affected child and together with disturbed sleep pattern may interfere with normal growth and development. Early diagnosis and treatment of leg pains is therefore a very important part of the management of VCFS patients.

As improvement in response to podiatric treatment was noticeable, biomechanical foot abnormalities must play a major role in the aetiology of leg pains in VCFS patients. This improvement was only partial however, rendering it likely that other factors are involved in the pathogenesis of leg pains in VCFS. In June 1997, based on this preliminary study and 2 surveys (unpublished data)²⁰, the VCFS Educational Foundation added two new features to the list of the recognised clinical features of VCFS. These are Chronic Leg Pains and Excessive Subtalar Joint Pronation.

Conclusion

The findings of this study are preliminary and are subject to further scrutiny. The aim at this stage is to attract the attention to this new feature and to suggest a non invasive method of treatment. It is recommended that the current treatment of leg pains in VCFS should consist of a simple insole. The type and the thickness of the insole should be the same as what would normally be used for the diagnosed foot biomechanical abnormality. The prescribed insole should provide extra cushioning by the use of a 3 mm poron base. Other podiatric measures, like footwear assessment and advice and muscle stretching exercises should not be omitted.

Research is currently underway to refine, follow up and add to the data of this preliminary study and to investigate some of the other potential mechanisms involved in the aetiology of leg pains in VCFS.

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Velocardiofacial Syndrome

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