Original Research

CLUBFOOT DEFORMITY IN DOWN'S SYNDROME

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ABSTRACT

The association of Down's syndrome (trisomy 21) with clubfeet has not previously been elaborated. Eight patients with a total of 15 clubfeet were identified for review. Five of these had trisomy 21 noted by chromosomal analysis, and 1 had a mosaic pattern. Two patients did not have chromosomal documentation, but had characteristic features of Down's syndrome. Interestingly, 2 patients had evidence of arthrogryposis as well as Down's syndrome. Four of the 8 patients had other orthopedic anomalies, including scoliosis, atlantoaxial instability, brachydactyly, and coxa valga. All 8 patients had an initial period of casting prior to any surgical intervention. Fourteen of the 15 feet required surgical intervention to afford correction of the deformity. The 6 feet with relatively long-term follow up (average: 5 years) showed that there was 1 excellent, 4 good, and 1 fair result using the criteria of Turco. It appears that, even though Down's syndrome is usually characterized by ligamentous laxity, when clubfeet are associated with this syndrome they are often resistant to non-operative treatment, and surgical treatment seems to produce an acceptable result.

Among the medical aspects of Down's syndrome, a number of orthopedic manifestations have been described. These include severe pes planus, atlantoaxial subluxation, patellar subluxation or dislocation, metatarsus varus, scoliosis, slipped capital femoral epiphysis, and subluxing and dislocating hips. Patients with Down's syndrome have been characterized as having generalized ligamentous laxity. However, we recently saw and treated a patient with Down's syndrome and clubfeet at this institution. A review of the literature revealed only a single passing reference to this condition in this group of patients (without any clinical specifics).

We therefore reviewed the records of all Down's syndrome patients treated at the Shriners Hospital for Crippled Children, Chicago Unit, for additional clubfoot patients to possibly elucidate the natural history and evaluate the results of treatment of this deformity in this subset of patients.

METHODS

The charts of all patients treated at the Shriners Hospital for Crippled Children, Chicago Unit, were reviewed, and 98 patients with the diagnosis of Down's syndrome were identified. Because of the referral nature and specialization at this institution, these patients may not be representative of the general population of Down's syndrome. We identified seven patients with a total of 13 clubfeet. One additional patient from the private practice of one of the authors who also manifested these combined deformities was found. We therefore had a total of 8 patients with a total of 15 clubfeet.

The medical records were reviewed with particular attention to chromosomal diagnosis, family history, orthopedic anomalies, operative and non-operative treatment of the deformities, and clinical follow up. All radiographs were reviewed, noting preoperative and postoperative measurements for the lateral talocalcaneal, anteroposterior (AP) talocalcaneal, and talo-first metatarsal angles.

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RESULTS

Of the 8 patients, 5 were identified as having trisomy 21 by chromosomal analysis, and 1 had a mosaic pattern. Two patients did not have chromosomal documentation available, but manifested the characteristic facies and other clinical stigmata of the syndrome. Seven of the 8 had bilateral foot involvement. One patient had a family history of clubfoot. Six other patients specifically denied any history of Down's syndrome or clubfeet. The family history of another was unknown. None of the patients had a history of birth trauma, and perinatal complications were confined to 1 patient born 5 weeks premature secondary to premature rupture of membranes.

Four of the 8 were free of other significant orthopedic anomalies. One patient had a mild scoliotic deformity and atlantoaxial instability. Another patient had bilateral brachydactyly and coxa valga. Two other patients were felt to be arthrogrypotic in addition to their main diagnoses. These patients had multiple joint contractions, including knees, elbows, and hips. One of these patients had trisomy 21 noted on chromosomal analysis, but the other was 1 of the 2 study patients who had not had a karyotype.

All eight patients had an initial trial of serial casting, and some had other additional closed methods prior to surgical intervention. The serial casting was done for an average of 5 months (range: 3 to 10). Some patients were maintained in corrective shoes for a period following casting. A wide variation in closed treatment was observed, as this was usually accomplished at an outside institution prior to referral to our hospital.

Four patients (6 feet) underwent surgical procedures to correct foot deformities prior to initial presentation. In 2 cases (2 feet) these consisted of tendo-Achilles lengthenings, and in 2 cases (4 feet), more extensive clubfoot releases were performed. Four patients had been managed solely by non-operative means prior to initial presentation.

The mean age at the time of our index procedure was 2 + 7 years (range: 0 + 7 to 11 + 2). Eleven feet required complete posterior–medial-lateral releases (four of these feet had undergone previous surgery). Two feet required limited releases (one foot with a previous surgical intervention). Two feet were managed non-operatively (one foot with previous surgical release).

For the 12 feet with adequate radiographic documentation, the mean AP talocalcaneal angle was 16° preoperatively and 26° postoperatively. The mean lateral talocalcaneal angle was 20° preoperatively and 31° postoperatively. The mean talo-first metatarsal angle was + 48° preoperatively and + 19° postoperatively, as measured by Simons.5

Follow up was variable, as the cases spanned a 20-year period, but generally consisted of serial clinical and radiographic evaluations. Excluding patients lost to follow up (1 foot), follow up that was too short (6 feet, less than 2 years since surgery), or inadequate documentation (2 feet), there were only 6 feet that could be classified after the criteria of Turco.6 The average length of follow up for these 6 feet was 64 months (range: 32 to 78). There was 1 excellent, 4 good, and 1 fair result. One foot required a supramalleolar rotational osteotomy due to persistent rotational deformity.

For the six feet with less than 2 years follow up, the average follow up was 5 months. They appeared to be doing well clinically and radiographically at their most recent evaluation. The patient who was felt to be arthrogrypotic but did not have chromosomal documentation was in this group (Table).

CASE EXAMPLE

A 2 + 0-year-old boy presented for evaluation. He was born at full term after a normal pregnancy, labor, and delivery. Birth weight was 7 lbs 1 oz. His family history is notable for a maternal uncle and first cousin with severe clubfeet. He was noted at birth to have characteristics suspicious for Down's syndrome, and a karyotype documented that he had trisomy 21. He was placed in serial casts at 3 days of age at an outside institution. These were changed until 3 months of age, at which time he was changed to prescription shoes for 1 year.

At his initial presentation, he was noted to have bilateral equinovalvar deformities with 10° to 15° of hindfoot varus, no active eversion, and passive dorsiflexion just to neutral. He underwent bilateral posterior medial-lateral releases at age 2 + 2 years. Postoperatively, he was casted for 6 weeks and braced for 9 months. At age 7 + 9 years, he required a left supramalleolar osteotomy to correct residual internal rotational deformity. Clinical follow up 78 months postoperatively revealed him to have ankle dorsiflexion to 15° bilaterally with mild residual metatarsus adductus that was passively correctable and with slight hindfoot valgus (Figs 1A-B).

DISCUSSION

The association of Down's syndrome with orthopedic deformities consistent with ligament and joint laxity is well known. This report of eight patients with Down's syndrome and clubfeet is, to our knowledge, the only description of a series of patients with these combined conditions, and the combination of these two problems appears improbable. On the one hand,
### Table

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<thead>
<tr>
<th>Pt</th>
<th>Down's Dx</th>
<th>Foot Involvement</th>
<th>Family History</th>
<th>C-Spine Stability</th>
<th>Other Ortho Conditions</th>
<th>Non-operative Treatment</th>
<th>Foot Surgery Elsewhere</th>
<th>SHCC Surgery</th>
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<td>Down's CF</td>
<td>N1</td>
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<td>L Talar</td>
<td>B PMLR</td>
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<td>Down's</td>
<td>Min Cl-2 Instability</td>
<td>Mild Scoliosis Min Cl-2 Instability</td>
<td>B Casts</td>
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<td>L Plantar Fasciotomy &amp; Abd Hall</td>
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<td>F/U Too Short</td>
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<td>Down's CF</td>
<td>NL</td>
<td>? Anthrogypotic B Knee FC</td>
<td>B Clubfoot Release 6 mos</td>
<td>B PMLR</td>
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*FC = Flexion contracture
CF = clubfoot
PMLR = posteromedial lateral release
SMO = supramalleolar osteotomy*

Down's syndrome and its ligamentous laxity would seem to defy the development of clubfeet. Yet these 15 feet were found to have all the characteristics of clubfeet, and all but one required surgical treatment to achieve deformity correction. Upon review, two patients—one with confirmed chromosomal abnormalities characteristic of Down's syndrome—and were felt to have signs and symptoms of an arthrogypotic nature. We were unable to find any reference in the literature to such a combination of conditions. Could it be that those patients with Down's syndrome who develop clubfeet have a localized involvement with arthrogyposis?

It is impossible to make statistically valid conclusions from a retrospective collection of a limited number of patients such as this, and we want to make absolutely clear that we do not purport this to be an outcome-of-treatment study or an incidence study of the clubfoot deformity in Down's syndrome patients. Additionally, this report cannot be construed as a natural-history study of untreated clubfeet in Down's syndrome; however, it does report what has happened to the clubfeet of this small group of patients over the short term. However, several observations can be made: this improbable combination of Down's syndrome and clubfoot does exist. Secondly, 14 of the 15 feet eventually required surgical intervention. Although there was a certain element of preselection prior to presentation at this institution, it appears as though this subset of clubfoot patients is as resistant to non-operative treatment and as likely to require surgical correction as the average clubfoot patient. The deformities in the patients able to be evaluated with adequate clinical and radiographic follow up tended to do well after an average follow up of 5 years. Unfortunately, only 6 of the 15 feet were in this category.

This study cannot determine whether or not there is a tendency for overcorrection in these patients over time, but perhaps one could speculate that the joint and ligament tightness condition, clubfoot, may prevent the overcorrection that one might expect in a ligamentously lax condition, Down's syndrome. Our results, such as they are, seem to indicate that the clubfoot deformity in Down's syndrome is amenable to surgical correction.
REFERENCES


EDITORIAL DISCUSSION

ORTHOPEDICS: Should the two patients who have not had a karyotype been included in your analysis of children with Down’s syndrome, as there is no proof that they have Down’s syndrome?

Miller et al: It is true that there is no chromosomal documentation. However, these patients exhibited all of the characteristics of Down’s syndrome patients. The children who had chromosome analysis did not demonstrate the characteristic features of Down’s syndrome. Interestingly, we describe a couple of patients who had arthrogrypotic-like findings in addition to the usual findings of Down’s syndrome. One of those patients had chromosome analysis that confirmed that he had Down’s syndrome; the other did not. If both of the patients with arthrogrypotic-like findings were the two without chromosome analysis, then it is clear that those two patients should have been eliminated completely from the study. Those two patients without chromosome analysis were patients identified during the period of time in which chromosome analysis was not routinely done at this institution, but the features and characteristics of Down’s syndrome patients were well known.

The purpose of our article was not to provide an all-inclusive dissertation on clubfeet in Down’s syndrome patients. As mentioned in the introduction, caring for a patient with this constellation of findings stimulated us to search the literature to find other reports on this association, and we were able to find none except for a passing reference, which we have listed. Therefore, we thought it worthwhile to alert the orthopedic community that this combination of Down’s syndrome with clubfoot does occur, and that in our selected series of patients, non-operative treatment was incapable of producing acceptable correction of the deformity. This article may serve as a stimulus for others who have a collection of these patients to evaluate their results, which perhaps are longer term than ours, or greater in number.