The orthopaedic manifestations and management of children with Stüve-Wiedemann syndrome

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Stüve-Wiedemann syndrome is an autosomal-recessive disorder characterised by bowing of the long bones, progressive scoliosis, episodic hyperthermia and respiratory distress, usually resulting in death in infancy. We reviewed five children with the condition who had been followed since birth and who survived into childhood with a mean age at operation of 7.8 years (5 to 14). There was marked functional impairment with dysplasia of the long bones and scoliosis. Treatment of the triplanar deformities of the femora involved the use of the Ilizarov technique with the Taylor Spatial Frame.

Walking was preserved and improved in three children along with considerable enhancement of the appearance. Early insertion of a growing rod to control the progressive juvenile scoliosis was beneficial. The use of the Taylor Spatial Frame is strongly recommended to address the major complex deformities of the lower limbs which are encountered in this condition and to prevent their progression.

Case reports

We describe five children, two sets of siblings with a mean age at operation of 7.8 years (5 to 14) with Stüve-Wiedemann syndrome who survived into childhood. In four the ability to walk has been maintained by the correction of severe long-bone deformity. The fifth child has yet to have her limb-length discrepancy corrected.

The facial features of Stüve-Wiedemann syndrome are characterised by a consistent pursed appearance of the mouth which was seen in all the five children (Fig. 1a). They also had varying degrees of camptodactyly (Fig. 1b), but had very functional hands. Their radii and ulnae were bowed, but to a much lesser extent than the long bones of the lower limb (Fig. 2). The elbows also had limitation of full extension of 10° to 15°. The hip, knee and ankle joints had normal movements and these were maintained post-operatively. All the children were in the 10th to 20th growth percentile and of short stature.

All children had a characteristic absence of fungiform papillae, giving the tongue a smooth appearance. They also had poor dentition with caries. Two (cases 1 and 2) had sustained two fractures each of their tibiae, all of which united uneventfully after application of a cast.

All the children were the offspring of first-cousin marriages and had dysautonomic symptoms with insensitivity to pain and episodes of instability of temperature. Each child had chronic corneal abrasions and scarring requiring ongoing ophthalmological care. Weight-bearing was grossly abnormal with precarious balance because of severe malalignment of the lower limbs (Figs 2b and 3). Walking within the home was possible, but the children required a wheelchair when outside.

There was marked varus of the femora and tibiae, associated with an abnormal cortical trabecular pattern and coxa vara (Fig. 2, Table 1).
All the children had progressive scoliosis and two underwent stabilisation with a growing rod. The deformities of the lower limbs included coxa vara (mean 92°, 80° to 100°), femoral and tibial intorsion, femoral genu varum (mean 18°, 10° to 25°), tibial genu varum (mean 29°, 20° to 35°), genu recurvatum (mean 12°, 10° to 15°), external femoral torsion (mean 46°, 40 to 50°) and tibial intorsion (mean 58°, 50° to 70°) (Table I). These deformities were progressive, resulting in bizarre gait patterns (Fig. 3). They could all stand and walk short distances. Orthopaedic intervention to realign the lower limbs was successful in producing a better balance and gait in four of the children. The fifth (case 5) is currently awaiting correction of her deformities. All the children became community walkers post-operatively with fewer falls and had a limited ability to run.

Management of musculoskeletal deformities. Multiple osteotomies in three planes were required in both the tibiae and the femora, as well as limb-length equalisation (Table II).

Patients one and two, the eldest in our series, had marked coxa vara which resembled a shepherd's-crook deformity similar to that seen in severe fibrous dysplasia (Fig. 2a). Both required a valgus osteotomy of the proximal femur (Table II).

All the five children had progressive varus, rotation and limb-length inequality of both tibiae and femora. It was therefore thought that the best method for dealing with these multiplane deformities was the Ilizarov technique using the Taylor Spatial Frame (Smith & Nephew Inc., London, United Kingdom), which allowed the necessary corrective manoeuvres to be made (Fig. 4).

This was undertaken in three patients (cases 1 to 3), one case (patient three) being treated elsewhere. We have also recommended this treatment for the remaining two children. One (case four) has had osteotomies of the tibia and the femur with plate and rod fixation but requires further aggressive correction.

The results of external fixation were excellent and its use was well tolerated by the children. Complications were minimal with only two pins requiring change for pin-track infection. All the osteotomies healed. There was delayed union of the tibia in one patient (case one). All the children walked assisted in their frames. The Taylor Spatial Frame was removed after ten to 12 weeks and
a long-leg cast was applied for four to six weeks and correction has maintained been to date.

Scoliosis is relentlessly progressive in children with Stüve-Wiedemann syndrome. Although we attempted bracing in two children (cases 1 and 3) it continued to progress. In both children it is currently in excess of 50° and will require surgical intervention.

**Discussion**

The patients reported initially by Stüve and Wiedemann\(^3,4\) were diagnosed as having a Schwartz-Jampel type-2 syndrome as were two of our patients (cases one and two). Most authors, including ourselves now believe that both of these conditions are a single entity.\(^12^-18\)

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**Table I. Details of pre- and post-operative musculoskeletal deformities (*) in the five patients**

<table>
<thead>
<tr>
<th>Deformity</th>
<th>Case 1 MS</th>
<th>Case 2 AS</th>
<th>Case 3 SR</th>
<th>Case 4 FR</th>
<th>Case 5 AA</th>
</tr>
</thead>
<tbody>
<tr>
<td>Scoliosis</td>
<td>Pre-operative</td>
<td>35 Thoracic</td>
<td>30 Thoracic</td>
<td>45/45</td>
<td>62 Thoracolumbar</td>
</tr>
<tr>
<td>Post-operative</td>
<td>N/A*</td>
<td>N/A</td>
<td>35/30</td>
<td>40</td>
<td>N/A</td>
</tr>
<tr>
<td>Genu recurvatum</td>
<td>Pre-operative</td>
<td>12</td>
<td>15</td>
<td>10</td>
<td>12</td>
</tr>
<tr>
<td>Post-operative</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Tibial genu varum</td>
<td>Pre-operative</td>
<td>30</td>
<td>35</td>
<td>30</td>
<td>30</td>
</tr>
<tr>
<td>Post-operative</td>
<td>-5</td>
<td>-5</td>
<td>0</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Femoral genu varum</td>
<td>Pre-operative</td>
<td>20</td>
<td>25</td>
<td>20</td>
<td>15</td>
</tr>
<tr>
<td>Post-operative</td>
<td>0</td>
<td>0</td>
<td>-5</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>External rotation femur</td>
<td>Pre-operative</td>
<td>45</td>
<td>50</td>
<td>45</td>
<td>50</td>
</tr>
<tr>
<td>Post-operative</td>
<td>10</td>
<td>10</td>
<td>5</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Tibial intorsion</td>
<td>Pre-operative</td>
<td>60</td>
<td>60</td>
<td>70</td>
<td>50</td>
</tr>
<tr>
<td>Post-operative</td>
<td>20</td>
<td>15</td>
<td>15</td>
<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>

* N/A, not available

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Case 1 – Photographs showing a) the pre-operative appearance of the bowed lower limbs at the age of six years with marked varus and limb-length inequality and b) the standing posture with typical hypereextension of the knees and precarious balance.

Fig. 3a

Fig. 3b
The geographical distribution of children with Stüve-Wiedemann syndrome appears to be concentrated in the Middle East with most cases being reported from Oman, Yemen, Sudan, and the United Arab Emirates. Although most children with the condition die in infancy, more reports of survival into childhood are appearing, including our case series.

Although the predisposition to malignant hyperthermia has always been an anaesthetic concern, it is noteworthy that our five children have had a total of 26 general anaesthetics to date with no temperature crisis intra-operatively. A recent report addressing the association of malignant hyperthermia and Stüve-Wiedemann syndrome concluded that the type of hyperthermia seen in this syndrome is not life-threatening malignant hyperthermia and that sevoflurane can be used safely in these children. Our experience supports this observation.

A molecular genetic study indicated that Stüve-Wiedemann syndrome is caused by a mutation in the leukaemia inhibitory factor receptor gene. This factor is a polyfunctional cytokine which affects the differentiation, survival and proliferation of a wide variety of embryonic cells. The gene map locus is chromosome 5p13.1 at marker D5S418. It is essential that the orthopaedic surgeon is involved early in the management of these children as their deformities are progressive and their chances of survival are improving. Our involvement was late in four of the children who developed major deformities before an orthopaedic opinion was sought. Although three of the children had undergone some correction by standard osteotomies with plate fixation or external fixators, the deformities recurred because correction had not been achieved in all planes. In view of the considerable three-plane deformity it is safer and more efficient to correct them slowly using the Ilizarov technique with the Taylor Spatial Frame.

Although limb-length inequality could readily be accomplished during the correction of the deformities because the mean amount of discrepancy was only 2 cm, Children with Stüve-Wiedemann syndrome also have decreased sensitivity to pain and therefore tolerate the pins and frame well. We found that they walked earlier with the

**Table II. Details of the surgical management in the five patients**

<table>
<thead>
<tr>
<th>Case</th>
<th>1 MS</th>
<th>2 AS</th>
<th>3 SR</th>
<th>4 FR</th>
<th>5 AA</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Scoliosis</strong></td>
<td>Bracing with thoracolumbar-sacral orthosis</td>
<td>Bracing with thoracolumbar-sacral orthosis</td>
<td>Growing rod</td>
<td>Growing rod</td>
<td>Bracing with thoracolumbar-sacral orthosis</td>
</tr>
<tr>
<td><strong>Coxa vara (°)</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pre-operative</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Post-operative</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Number of osteotomies of the femur</strong></td>
<td>1 (age 5) ex fix</td>
<td>1 (age 9) ex fix</td>
<td>1 (age 3) plate</td>
<td>1 (age 5) Ilizarov</td>
<td>Nil</td>
</tr>
<tr>
<td></td>
<td>2 (age 8) TSF</td>
<td>2 (age 12) TSF</td>
<td>1 (age 5) Ilizarov</td>
<td></td>
<td>Nil</td>
</tr>
<tr>
<td><strong>Number of osteotomies of the tibia</strong></td>
<td>2 (age 5) ex fix</td>
<td>2 (age 9) rod</td>
<td>3 (age 3) rodding</td>
<td>1 (age 3) plate</td>
<td>Nil</td>
</tr>
<tr>
<td></td>
<td>1 (age 8) TSF</td>
<td>1 (age 12) TSF</td>
<td>1 (age 5) TSF</td>
<td></td>
<td>Nil</td>
</tr>
<tr>
<td><strong>Lengthening of the tibia (cm)</strong></td>
<td>2 (age 8) (TSF)</td>
<td>3 (age 12) (TSF)</td>
<td>Nil</td>
<td>Nil</td>
<td>Nil</td>
</tr>
<tr>
<td><strong>Lengthening of the femur (cm)</strong></td>
<td>2 (age 8) (TSF)</td>
<td>Nil</td>
<td>1 (age 3) (Ilizarov)</td>
<td>Nil</td>
<td>Nil</td>
</tr>
</tbody>
</table>

*TSF, Taylor Spatial Frame*
frames, which encouraged healing of the osteotomies and that they tolerated removal of the frame and the pins as outpatients.

We staged the operations by correcting the tibia and the femur in one limb and then proceeding to the other limb when healing and return of joint movement was complete. Once the scoliosis has exceeded a 40° Cobb angle, we recommend the insertion of a growing rod because the curve progresses inevitably in spite of orthotic management.

No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.

References