The long-term function of the knee in patients with fibular hemimelia and anterior cruciate ligament deficiency

Most patients (95%) with fibular hemimelia have an absent anterior cruciate ligament (ACL). The purpose of this study was to assess the long-term outcome of such patients with respect to pain and knee function. We performed a retrospective review of patients with fibular hemimelia and associated ACL deficiency previously treated at our institution. Of a possible 66 patients, 23 were sent the Musculoskeletal Outcomes Data Evaluation and Management System (MODEMS) questionnaire and Lysholm knee score to complete. In all, 11 patients completed the MODEMS and nine completed the Lysholm score questionnaire. Their mean age was 37 years (27 to 57) at review. Five patients had undergone an ipsilateral Symes amputation. There was no significant difference in any subsections of the Short-Form 36 scores of our patients compared with age-matched controls. The mean Lysholm knee score was 90.2 (82 to 100). A slight limp was reported in six patients. No patients had episodes of locking of the knee or required a supportive device for walking. Four had occasional instability with sporting activities.

These results suggest that patients with fibular hemimelia and ACL deficiency can live active lives with a similar health status to age-matched controls.
where 100 represents an absence of pain. Additional questions sent to the patients included information relating to their current occupation, hobbies and further surgical history since discharge from our institution. Patients who completed the MODEMS questionnaire were also asked to complete a Tegner and Lysholm knee score, which is a validated outcome measure to assess knee pain, instability and lower limb function.

Data analysis on the SF-36 component of the questionnaire was compared with both a normal age-matched population as well as with a group of patients with traumatic ACL rupture prior to reconstruction. Statistical analysis. Unpaired t-tests were used to determine significance between each tested variable. Significance was set at p < 0.05.

### Results

A total of 66 patients were identified with fibular hemimelia and ACL deficiency. Of these, one had died, another had a mental disorder preventing participation and 41 patients were lost to follow-up or were uncontactable. The remaining 23 patients were traced and sent surveys, 14 of which were returned. Of these, two completed surveys were discarded due to prior ipsilateral above-knee amputation and another was withdrawn because of previous arthrodesis of the knee, leaving 11 patients for assessment, who formed this case series. One survey was only partly completed for the hip and knee pain score, so those data were excluded from analysis. Of the 11 patients who were sent the Lysholm Knee Score, nine completed it.

The mean age of the 11 patients at review was 37 years (27 to 57) of whom ten were in employment and most had active hobbies (Table II). None received social security or disability benefits, or were involved in a worker's compensation claim. The highest education level achieved was: high school in three, attended college in three, graduated college in two, and postgraduate degree in three. No patients reported a slight limp and four of these had undergone an ACL reconstruction for their instability.

### Discussion

We found that our patients with fibular hemimelia and ACL deficiency can live very functional lives. Many had physically demanding hobbies such as skiing, football and baseball. Occupations ranged from manual labour to office work with the latter group still involved in active recreation. Half of the patients obtained a college or postgraduate degree.

Our patients had very similar health to an age-matched population control group. We recognise that using the SF-36 as a tool to measure functional outcome has limitations. However, it does assess whether a patient is restricted in their work and activities or is affected by pain. The addition of a knee and hip pain scale as well as the Lysholm Knee Score specifically determined disability in the lower limb. We found no increase in pain in the limbs affected by fibular hemimelia compared with the contralateral limbs.
Those who had undergone a Symes amputation more often had a limp and had more difficulty squatting.

Instability of the knee following traumatic ACL rupture is common and usually necessitates reconstruction for return to sports which require pivoting of the knee. Mizuta et al.\textsuperscript{17} reviewed 18 patients under 15 years of age who were treated conservatively following traumatic ACL rupture. At a mean of 36 months follow-up only one patient had returned to their previous level of athletic activity and 13 (72\%) had instability of the knee daily as well as knee pain with exercise.\textsuperscript{17} These findings of instability after traumatic ACL rupture are also found in adult patients with up to 77\% reporting instability when following the natural history of ACL rupture.\textsuperscript{18} Our patients with congenital ACL deficiency had less instability. Our results are in agreement with those of other authors who have reported varying degrees of knee instability in patients with congenital absence of the cruciate ligaments. In a series of ten patients with congenital absence of the ACL, Thomas, Jackson and Aichroth\textsuperscript{11} found that 50\% reported weekly episodes of the knee giving way. Their

\begin{table}
\centering
\caption{Patient demographics}
\begin{tabular}{llll}
Patient number & Age at review (yrs) & Category & Occupation & Hobbies \\
1 & 29 & Ia & Housekeeper & Water sports, snow skiing, biking \\
2 & 45 & Ib & Engineer & Motorcycles, baseball, football \\
3 & 40 & Ia & Legal administrator & Volleyball, skiing \\
4 & 27 & II & Staff accountant & Golf, horseback riding \\
5 & 27 & Ia & Farmer & Hunting, fishing, golf \\
6 & 31 & Ia & Financial planning & Baseball, biking, camping, gardening \\
7 & 36 & II & US Army civilian & Sky diving, hang gliding \\
8 & 57 & II & Physician & Hunting, fishing, automobile racing \\
9 & 29 & II & Advertising/Marketing & Sports (specified) \\
10 & 34 & II & Operations manager & Bowling, travel \\
11 & 48 & II & Courier & None \\
\end{tabular}
\end{table}

\textsuperscript{*} Achterman and Kalamachi Classification\textsuperscript{1}

\begin{figure}
\centering
\includegraphics[width=\textwidth]{fig1}
\caption{Bar chart showing the mean Short-Form 36 (SF-36) scores in patients with fibular hemimelia compared with an age-matched population (whiskers indicate the standard deviation).}
\end{figure}
cohort had a spectrum of associated diagnoses including three patients with fibular dysplasia. Roux and Carlioz\textsuperscript{10} found a much lower incidence of problems with only 3% of their cohort of 69 patients with fibular hemimelia and absent cruciates reporting knee instability.

It is not clear why instability of the knee is not a more frequent symptom in patients with fibular hemimelia. Gabos, El Rassi and Pahys\textsuperscript{19} reported anatomical variations in patients with fibular hemimelia which might provide stability. In four patients with congenital ACL deficiency on whom they performed ACL reconstruction, they found a hypertrophied ligament of Humphries at arthroscopy in three patients. They hypothesised that the hypertrophied ligament might contribute to the stability of the knee. Clearly, other intra-articular structures might be recruited as secondary stabilisers when the ACL is absent or non-functional.\textsuperscript{19}

Other authors have suggested that patients with congenital absence of the cruciate ligaments "adapt to their instability".\textsuperscript{20} Although this may be true, none of our patients indicated that they avoided specific activities but they might have modified their activities to suit their circumstances. Every patient places different demands on their knee and,

\begin{table}[h]
\centering
\begin{tabular}{|c|c|c|c|}
\hline
SF-36 sub group & Fibular hemimelia & Traumatic ACL\textsuperscript{*} & p-value\textsuperscript{†} \\
\hline
Physical function & 89.6 (15.7) & 85.2 (13.5) & 0.3 \\
Role physical & 88.6 (25.9) & 60.1 (38.4) & 0.018 \\
Bodily pain & 70.7 (20.9) & 77.8 (21.8) & 0.3 \\
General health & 72.8 (20.9) & 80.7 (12.1) & 0.042 \\
Vitality & 55.5 (21.5) & 73.6 (46.7) & 0.2 \\
Social functioning & 80.7 (27.0) & 78.9 (20.8) & 0.8 \\
Role emotional & 84.9 (34.5) & 81.2 (31.6) & 0.74 \\
Mental health & 70.9 (23.1) & 77.1 (15.0) & 0.2 \\
\hline
\end{tabular}
\caption{Mean (SD) Short-Form 36 (SF-36) scores in patients with fibular hemimelia compared with patients with traumatic anterior cruciate ligament (ACL) disruption}
\label{tab:36}
\end{table}
as our data supports, some patients with congenital ACL deficiency have symptoms of instability of the knee whereas others may not. For those patients with congenital ACL deficiency who have instability, Gabos et al\textsuperscript{19} found that ACL reconstruction with a transphyseal approach for allograft reconstruction can restore stability and function.

There are several limitations to this study. Most important was our low response rate with only 11 of 66 patients completing the MODEMS and nine of 11 completing the Lysholm score. We should be careful to draw broad conclusions about patients with fibular hemimelia when our cohort only represented one-sixth of the patients that met the inclusion criteria. Furthermore, we did not have MRI or arthroscopic images of our patients’ knees to confirm ACL deficiency. Lastly, we did not evaluate our patients clinically to objectively document the stability of the knee or the findings on examination of the lower limb. Most patients had moved away from our institution and examination would not have been feasible.

Our cohort of patients with fibular hemimelia and ACL deficiency demonstrated that many can live very active lives similar to a health and age-matched population. In our limited review of a total of 66 patients, amongst those 11 we were able to trace, most did not have any instability or pain in the knee.

**Supplementary material**

A table showing the Lysholm scores for all patients is available with the electronic version of this article on our website www.jbjs.org.uk

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**Table IV. Hip and knee scores in patients with associated classification of fibular hemimelia and previous associated surgery**

<table>
<thead>
<tr>
<th>Patient number</th>
<th>Type of fibular hemimelia*</th>
<th>Previous surgical procedure</th>
<th>Ipsilateral knee pain score</th>
<th>Ipsilateral hip pain score</th>
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</thead>
<tbody>
<tr>
<td>1</td>
<td>Ia</td>
<td>None</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>2</td>
<td>Ib</td>
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<td>87</td>
<td>93</td>
</tr>
<tr>
<td>3</td>
<td>Ia</td>
<td>Contralateral epiphysodesis</td>
<td>80</td>
<td>87</td>
</tr>
<tr>
<td>4</td>
<td>II</td>
<td>Symes</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
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<td>Ia</td>
<td>Contralateral epiphysodesis</td>
<td>100</td>
<td>100</td>
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<td>Ia</td>
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<td>100</td>
<td>100</td>
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<td>100</td>
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<td>II</td>
<td>Symes</td>
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<td>100</td>
</tr>
<tr>
<td>11</td>
<td>II</td>
<td>Symes</td>
<td>-</td>
<td>-</td>
</tr>
</tbody>
</table>

* Achterman and Kalamachi Classification\textsuperscript{1}
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