CLINICAL PATTERNS OF PRESENTATION OF LIMB DEFICIENCY IN CHILDREN IN THE ROEHAMPTON CLINIC
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INTRODUCTION
Limb deficiency is rare in children and varies from almost undetectable to complete absence and those with sufficient severity thought to warrant prosthetic prescriptions are referred to the rehabilitation clinics. Number of cases with longitudinal and distal transverse deficiencies are managed by paediatric orthopaedic and plastic surgical colleagues with interest in management of limb malformations. The initial care is directed not just at the index child but towards parents, siblings, carers, and school staff. It is vital the child has access to all the professional skills needed to achieve the optimal result. Specialist staff involved - paediatric, orthopaedic, plastic surgical, rehabilitation - should communicate freely and form a coordinated plan of management for the individual child according to the clinical and social background, avoiding conflict of advice and inappropriate timing of treatment. The parents are invariably and understandably anxious and concerned and should form part of the multidisciplinary structure. Management of the limb deficient child whatever the cause, differs in many aspects from that of the adult amputee, (1). This study analyses the varying clinical presentations of limb deficiencies in children attending Roehampton Clinic in preparation to study their pattern of prosthetic use.

INCIDENCE AND PREVALENCE
The incidence and prevalence of limb deficiency varies throughout the world depending on the criteria used to collect the information. Most studies suggest an incidence of 50/100,000 (2). In the UK the incidence of significant limb deficiency requiring prosthetic assessment is approximately 12.5/100,000 live births and occurred most commonly in the age group 0-9 years (10/21) but from trauma in the age group 10-49 years (33/54), (3) For the total population the prevalence of limb deficiency in the UK is 132/100,000, (4). The Roehampton prosthetic clinic primarily serves the South West Thames Health region with a population of 3.2 million. The total population of amputees attending Roehampton is 3,200. Patients living in the borderline districts attend clinics in the neighbouring regions. There were 260 patients identified in the age group 18 years and below who attended Roehampton during the last 5 years. Though epidemiological data of distribution of limb deficiencies in different age groups in the various health regions in the UK are not available for comparison, there appears to be a higher proportion of children attending Roehampton. This is explained by the historical national role of Roehampton in this field attracting tertiary referrals of children specially with multi limb deficiencies from other regions.

* Full manuscript not available at time of publishing.
SITES AND LEVELS

There were 129 male and 131 female children in the series. 79.6% (207/260) children had single limb deficiency (table 1). The 260 patients had 346 limbs affected with varying degrees of deficiencies and handicap. The pattern of distribution among the four limbs is shown in table 2. As widely recognised the upper limbs are more frequently affected (69.6%).

In the past various combinations of Greek and Latin terms have been used to describe limb anomalies resulting in difficulties to compare data. Use of several synonyms to describe a congenital anomaly has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion. The distribution of different upper and lower limb malformation has added confusion.

It is therefore vital to record parental age, consanguinity, number of siblings and state of health (7, 8, 9).

In this series of 260 patients 70% (181/260) of patients had limb deficiency as an isolated congenital anomaly with no identifiable cause. Another 20 patients had associated non-limb defects though could not be identified as a syndrome. 52 patients in the series could be categorised into a recognisable syndrome. It must be noted that some syndromes of limb deficiency are mere collection of heterogenous conditions with no plausible genetic transmission.

MEC'93

Transverse deficiencies in the upper limb formed 82.9% (200/241) of which the partial forearm (below elbow) accounted for 50.5% (101/200). It is unusual for the partial forearm (below elbow) deficiencies to be associated with other congenital defects. Of the 101 patients all but nine had classical upper third level deficiency. A predominant female and left sided incidence has been recognised in other studies (5). In this survey there were 57.4% females and 62.6% incidence on the left.

Carpal, metacarpal and phalangeal deficiencies formed 33.6% (8/241) of the upper limb deficiencies. This is a relatively high percentage seen in a prosthetic rehabilitation clinic and is due to the combined approach of management of these children together with the hand surgeons.

The upper arm partial (above elbow) level in this series (8/11) was found to be part of multi-limb involvement. One of the patients with single limb involvement at this level had a solitary terminal vestigial digit which needed surgical removal to avoid pressure from the prosthetic socket.

Transverse upper arm total (through shoulder) presented usually in a bilateral fashion and unlike in Thalidomide induced limb reduction defects these children showed normal shoulder contours. The glenoid process was at least partially present (6).

Most of the pre axial (radial) longitudinal defects were 20th bilateral, whereas a number of patients with longitudinal post axial (ulnar) defects had involvement of the lower limbs. Malformations which have been referred to as femur-fibula-ulna (FFU) syndrome.

AETIOLOGY

In a substantial number of cases it is not possible to identify any genetic or exogenous causes for the limb deficiency. Knowledge of the early development of the limbs, studying patterns of abnormal embryogenesis and animal research has been helpful in understanding multiple congenital anomalies. It is important to give correct advice about management, prognosis and risk of recurrence. Limb defects can be due to single major gene showing typical mendelian pedigree pattern. Conditions such as cleft hand cleft foot syndrome and Holt-Oram show autosomal dominant pattern of inheritance whereas thrombocytopaenia absent radius (TAR) and Roberts syndrome are autosomal recessive. Chromosomal aberrations such as trisomy 13 and trisomy 21 are accompanied by polydactyly and brachydactyly respectively. Abnormality such as a ring chromosome 13 leads to absence of thumb. However, the pattern of inheritance is by no means clear cut and some genes may have variable effects. Consanguinity of the parents suggest autosomal recessive inheritance. Increased paternal age is associated with dominant mutations such as achondroplasia, Marfan's syndrome, and Apert syndrome. It is therefore vital to record parental age, consanguinity, number of siblings and state of health (7, 8, 9).

In this series of 260 patient 70% (181/260) of patients had limb deficiency as an isolated congenital anomaly with no identifiable cause. Another 20 patients had associated non-limb defects though could not be identified as a syndrome. 52 patients in the series could be categorised into a recognisable syndrome. It must be noted that some syndromes of limb deficiency are mere collection of heterogenous conditions with no plausible genetic transmission.
Traumatic cause of limb loss in children is fortunately rare. The types of trauma ranged from road traffic accidents, commercial food processing equipment to bite injury by a gorilla.

<table>
<thead>
<tr>
<th>Congenital Limb Defects (Isolated)</th>
<th>181</th>
<th>69.6%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital limb defects and other anomalies (Unrecognised)</td>
<td>20</td>
<td>7.7%</td>
</tr>
<tr>
<td>Congenital limb defects and other anomalies (Syndromes)</td>
<td>32</td>
<td>12.3%</td>
</tr>
<tr>
<td>Trauma</td>
<td>10</td>
<td>3.8%</td>
</tr>
<tr>
<td>Vascular</td>
<td>8</td>
<td>3.1%</td>
</tr>
<tr>
<td>Malignancy</td>
<td>7</td>
<td>2.7%</td>
</tr>
<tr>
<td>Infection</td>
<td>2</td>
<td>0.8%</td>
</tr>
</tbody>
</table>

Ablation of limbs for neoplasia is becoming rarer as a result of improved management of skeletal malignancies resulting from early diagnosis with the aid of CT and MRI scans. Limb conserving surgery with and improved success with chemo and radiotherapy. Ablation surgery is a viable treatment option for skeletal malignancies. Avoiding amputations endoprostheses is a viable treatment option for skeletal malignancies. The single upper limb amputation was at the shoulder level. Four of the six lower limb amputations were through hip or hind quarter amputations.

The vascular aetiology was the commonest in the adult population, contributing to this paediatric group for 8 (5.1%) amputations. A single case of Klippel-Trenaunay syndrome resulted in a through hip disarticulation, (10). Septicaemia due to streptococcal or meningococcal infections resulted in major amputations in three children, (11). Occasionally, limb loss occurs after essential vascular access.

Summary
The management of limb deficiency in children is a specialised aspect of rehabilitation and often involves a range of professional skills. Appropriate prosthetic management and often involves a range of professional skills. Appropriate prosthetic rehabilitation and often involves a range of professional skills. Appropriate prosthetic rehabilitation and often involves a range of professional skills. Appropriate prosthetic rehabilitation and often involves a range of professional skills.

REFERENCES
3. Dept. of Health *Amputation Statistics for England Wales and N Ireland 1989* *Statistics and Management Information Division, SC Fylde, Room 7116, North Fylde, Central Offices, Preston, FY6 8JA*