

7. CONGENITAL LIMB DEFICIENCY

- 7.1 The birth of a child with congenital abnormalities of the limbs is a cause of great anxiety to the parents and family. They require an adequate explanation, reassurance that experts are available to give them detailed advice regarding these rare conditions, practical assistance and counselling¹. Provided other life threatening congenital abnormalities are absent, these children are expected to develop normally in the early months and until they are much older and start to compare themselves with their peers, they will not have the sense of loss associated with acquired limb deficiency. Such children instinctively tend to use the limbs they have to interact with the environment and to mobilise.
- 7.2 The management of these patients is effectively from birth throughout the individual's life, and involves various professional disciplines at different stages². It is made considerably more complex if more than one limb is deficient. In the early stages, it is primarily the parents who are being supported, with the emphasis shifting progressively to the child as he or she becomes older. Unfortunately, the non-registration rate for adults with congenital upper limb deficiency could be as high as 64 percent³.

ROLE OF DISTRICT GENERAL HOSPITAL

- 7.3 The infant should be seen in the neo-natal period by the paediatrician to recognise the limb deficiency, exclude other congenital anomalies, and give initial advice and information to the parents. It is often helpful for a locally based paediatrician to take on responsibility for the longer-term follow-up of the child.
- 7.4 Those infants with lower limb abnormalities where major joint involvement is present or suspected, must be seen at an early stage by an orthopaedic surgeon (preferably a paediatric orthopaedic surgeon), in particular to test the hip joints for subluxation or dislocation. In certain cases, for example proximal femoral focal deficiency, a paediatric orthopaedic surgeon should keep the child under long-term review. For most cases of congenital limb deficiency, early surgery on the extremity should be avoided. It is important to give time to see how the individual child develops, and for the parents to understand the benefits and limitations of surgery. Even in the minority of cases where there is an indication for surgery, it will usually be a treatment option, rather than a necessity.
- 7.5 A designated social worker (or depending on local arrangements another designated individual such as a therapist) should assist the family from an early stage, to provide general support and counselling, and to advise on benefit entitlements and the relevant voluntary support organisations. These comprise REACH for children with upper limb deficiency, and STEPS for children with lower limb deficiency.
- 7.6 The Consultant Paediatrician (or by local arrangement a designated consultant from another discipline) should refer the child to the appropriate Limb Deficiency Clinic as early as practicable, and ideally within the first month of life (unless this is inappropriate due to other life threatening problems). This is not because early treatment is usually necessary, but to ensure that the parents receive detailed and accurate specialist advice.
- 7.7 Although routine ultrasound scans still sometimes miss these abnormalities, if a limb deficiency is detected during pregnancy, the parents should be referred ante-natally to the appropriate Limb Deficiency Clinic.
- 7.8 Good liaison between the paediatric service, Limb Deficiency Clinic, and orthopaedic surgeons is vital. It should be borne in mind that the management of almost all children with transverse limb deficiencies is by provision of a prosthesis or advice, and surgery is rarely indicated. Children with longitudinal limb deficiencies may require both a prosthesis and surgery.

ROLE OF THE LIMB DEFICIENCY CLINIC (at PARC)

- 7.9 Depending upon the type of deficiency, the family may require only an advisory service. Various aids or 'gadgets' may be suggested, or a prosthesis or simpler custom-made appliance may be indicated. A minority will be helped by surgery. The optimal timing of prosthetic fitting and/or surgery should be discussed.
- 7.10 Ideally, the family should be seen at a special Limb Deficiency Clinic, where all the necessary expertise can be concentrated, and this will help to ensure that there is a critical mass of such patients to ensure optimal levels of care⁴. This Clinic also provides parents with an opportunity to meet other families with similarly affected children. Because many of these patients will require prostheses or similar appliances, a Tertiary Referral PARC is a suitable base for the Limb Deficiency Clinic. These conditions are rare, thus smaller PARCs are unlikely to have sufficient numbers of such patients mentioned above⁵. Ideally therefore, the care of these children should be concentrated at the larger Tertiary Referral PARC, at least initially. However, to take into account patients' wishes, problems of travelling and access etc, some of the more established patients, particularly those with relatively straightforward needs, may be seen at the local Prosthetic and Amputee Rehabilitation Centres, provided good clinical links are maintained with the larger Centre.
- 7.11 The clinical team at the Limb Deficiency Clinic should be led by a consultant who is a specialist with expertise in congenital limb deficiency, prosthetics, and Rehabilitation. Ideally the consultant should see the infant with his or her parents by about one month of age (certainly before 6 months). The limb deficiency should be classified using the ISO system⁶. This will allow the parents to be given more specialist advice on the prognosis for their child, and on the options available for suitable short and long-term management and Rehabilitation. The consultant at this Clinic should also be able to advise the parents and surgeons regarding possible reconstructive surgery, including the optimal timing of such surgery, if indicated, from the point of view of the child's overall development. For example Syme's amputation for a major longitudinal deficiency of the fibula is often appropriate shortly after the child is old enough to walk, and this would allow early fitting of an end bearing prosthesis⁷. However, close liaison with a specialist paediatric orthopaedic surgeon with experience of these uncommon children's conditions is vital, particularly in cases of rarer and more variable types of deficiency, such as proximal femoral focal deficiency⁸.
- 7.12 Patients with major limb deficiencies should remain under the care of the named consultant at the Clinic indefinitely, and after the initial medical referral, the family should be able to seek appointments at the Clinic directly (ie without having to be referred each time by their General Practitioner).
- 7.13 Involvement of a specialist occupational therapist at an early stage is essential for children with upper limb deficiency. The occupational therapist will initially advise the parents, and will subsequently supervise prosthetic training (if appropriate) together with one and two-handed activities, and provide advice and support when the child is starting at school.
- 7.14 Although most children with congenital lower limb deficiency, even those with secondary complications, will use their prosthesis for daily activities^{9,10}, and learn to walk on their own (with or without a prosthesis), those with more proximal loss or more complex disabilities will need the help of a specialist physiotherapist and in all cases parents should have access to one. Parents should have the option of seeing a counsellor with special experience of patients with limb deficiency, and should be given the names and addresses of voluntary organisations.

OTHER SPECIALISTS INVOLVED AT SPECIALIST LEVEL

- 7.15 **Medical Genetics:** Parents should be offered an appointment with a Consultant in Medical Genetics, to advise on the risk of congenital abnormalities in any future pregnancy, and in the offspring of the affected child.
- 7.16 **Orthopaedic Surgeon:** As mentioned above, close liaison with a specialist Paediatric Orthopaedic Surgeon is important in many of these cases. This is particularly important in cases where hip instability is present or suspected, and where either limb lengthening or amputation are options in treatment.
- 7.17 **Plastic Surgeon/Hand Surgeon:** For patients with partial deficiency of the hand, the advice of a hand or

plastic surgeon should be sought within the first 6 months of life, as more surgical options may be available at a younger age¹¹. This is particularly relevant in cases of syndactyly where this may require separation, or where for example pollicisation of an index finger or digital transfer, require consideration.

- 7.18 It is recommended that the Prosthetic and Amputee Rehabilitation Centre should have well-established links with these specialist surgeons, and joint clinics are an excellent way of bringing this about.

PROSTHETIC TREATMENT

- 7.19 Children who have an upper limb deficiency (such as a transverse deficiency of the radius and ulna partial), which is likely to be helped in later life by a prosthesis, should start using a simple cosmetic arm. Limb fitting should be undertaken when independent sitting balance is achieved at about 6 months^{12,13}. A functional body or electrically powered limb would generally be introduced at about 18 months of age, once the child is well established with walking, with more complicated control mechanisms being added later^{12,14}.
- 7.20 Patients requiring an upper limb prosthesis are a small group, and supervision of training in the use of artificial arms should be by a specialist occupational therapist based at the PARCs. This occupational therapist will also advise on the use of other appliances, aids or 'gadgets' and one-handed activities.
- 7.21 Children with lower limb deficiency should commence prosthetic fitting (and training) when they show signs of being ready to walk. Unilateral lower limb deficient children at any level and those with bilateral loss from below the knee are ready for prosthetic fitting when they pull to stand between 9 to 12 months^{15,16}. Unless an early amputation is appropriate, an extension prosthesis may be required, either of below knee end weight bearing or ischial bearing type, depending upon the degree of stability at the knee and hip joints.
- 7.22 Where possible, all children with either congenital or acquired limb loss should be treated by the same team of doctors, nurses, therapists, and prosthetists, to retain continuity, ensure a high level of expertise, and to provide an opportunity for families with similarly affected children to meet. New prostheses (or new sockets) should be delivered within two weeks. To facilitate this and to maximise the prosthetist's control over the manufacturing process, all prostheses should be fabricated at the PARCs. Delay in obtaining component parts for the prosthesis is liable to delay production; if such components are not rapidly and reliably available from the manufacturer or importer, then adequate stocks should be held. Ideally the prosthetists dealing with this group of patients should also be proficient in related orthotic treatment; failing that, ready access to a suitably experienced orthotist is essential. This could be by means of a joint clinic.
- 7.23 Some children will benefit from in-patient treatment at particular times, and facilities should be available to admit the child with a parent close to the Limbless Rehabilitation Centre.
- 7.24 Children using prostheses should be followed up by the Rehabilitation physician at 3-4 monthly intervals to allow alterations required by growth and changing needs to be made. Other patients should be able to make appointments easily and quickly when required.
- 7.25 The acceptance and usefulness of upper limb prostheses varies considerably between apparently similar individuals or levels of deficiency. The main disadvantage of a prosthesis is that it lacks sensation, which is a crucial part of normal hand function. Some individuals become very skilled in the use of their feet for prehension and should not be discouraged from doing so. Children should, however, be given the opportunity to try artificial arms¹². Except in cases of very high bilateral deficiency, lower limb prostheses are generally well tolerated and heavily used.

EDUCATION

- 7.26 Most children with congenital limb deficiencies are capable of attending a school, and should do so. Often a school visit by the specialist occupational therapist shortly before the child starts school, with a follow-up shortly thereafter, is very helpful in ensuring that the school staff understand, and can therefore best help the child, with or without a prosthesis. Children with more severe deficiencies may require some physical help (eg with negotiating stairs, use of the lavatory etc) but such assistance should be kept to the minimum and be as unobtrusive as possible. Some will require use of a wheelchair, which may pose problems of access.

PARC staff may need to participate in preparing a Statement of Educational Need.

ADOLESCENCE

- 7.27 Adolescents require particularly sensitive empathy as they become more concerned with their body image and relationships, and different strategies may need to be adopted for coping with everyday difficulties; for example, a child may always have had help from a parent with washing and dressing, but this may no longer be acceptable to a teenager. Specialist advice may be required in terms of considering a suitable career.

ADULT LIFE

- 7.28 In adult life, the prosthetic needs of many patients with congenital limb deficiency will continue to be more complex than those of people with acquired amputation, and they will continue to require the assistance of a specialist medical and prosthetic team. The vast majority of people with congenital limb deficiency have a normal life expectancy, but they may develop increased difficulties as they become older. For example, those with a lower limb deficiency may develop back pain due to premature secondary degenerative changes, and those with bilateral upper limb deficiencies who have used their feet for prehension may develop problems in the joints of their lower limbs as they become older.

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