Limb Deficiencies (Reduction Defects)

(Core Condition)

Description

Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), fingers (phalanges), thigh (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Inclusions

Transverse limb deficiency (reduction) – Complete or partial absence of the distal (furthest from the body) structures of the arm or leg in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. Selected terms used for types of transverse limb deficiencies include:

- Acheiria – Absence of a hand
- Adactyly – Absence of digits (fingers or toes), excluding isolated missing thumb (see below)
- Aphalangia – Absence of phalanges. Fingers contain 3 phalanges each. The thumb (pollex) and big toe (hallux) contain 2 phalanges. The other toes contain 3 phalanges each.
- Amelia – Complete absence of the upper limb (humerus, radius, ulna, wrist, hand and fingers) or complete absence of the lower limb (femur, tibia, fibula, ankle, foot, and toes).
- Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal deficiency (reduction).
- Oligodactyly – Deficiency of fewer than 5 digits.

Transverse terminal deficiency (reduction) – Complete absence of the distal structures of the arm with the proximal structures intact. This term usually refers to deficiency below the elbow, or complete absence of the distal structures of the leg with the proximal structures intact.

Congenital amputation, type not specified.

Longitudinal limb deficiency (reduction) – Partial absence of the upper limb in parallel with the long axis of the arm or partial absence of the lower limb in parallel with the long axis of the leg. These may involve preaxial (on the thumb side/ on the big toe side), postaxial (on the fifth finger side/ on the fifth toe side), or central parts of the arm or leg. Selected terms used for types of longitudinal limb reductions include:

- Ectrodactyly
- Ectromelia
- Isolated missing thumb
- Lobster claw hand
- Radial, ulnar, tibial, or fibular aplasia or hypoplasia
- Radial, ulnar, tibial, or fibular ray deficiency

Split-hand malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb deficiency (reduction) in which there is complete or partial absence of one or more of the central rays (second through fourth fingers and their associated metacarpal bones) of the hand.

	Split-foot malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb deficiency (reduction) in which there is complete or partial absence of one or more of the central rays (second through fourth toes and their associated metatarsal bones) of the foot. Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the upper limb or lower limb with all or part of the distal segment present. Phocomelia is a general term used for any type of intercalary limb reduction. Deficiency (reduction defect) of the upper limb or lower limb not elsewhere coded or of unspecified type – Complete or partial absence of the upper limb or lower limb that does not fall within the above categories or for which there is no specific description.
Exclusions	Shortened arms, forearms, hands, upper and/or lower legs, feet, toes or fingers that have all of their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism. Hypoplastic nails
ICD-9-CM Codes	755.2 – 755.4
ICD-10-CM Codes	Q71.0 – Q71.9, Q72.0 – Q72.9, Q73.0 – Q73.8
CDC/BPA Codes	755.20 – 755.49
Diagnostic Methods	Limb deficiencies (reductions) are usually easily recognized on physical examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:
The terminology for limb deficiency (reduction) is often confusing. Some terms (such as “phocomelia”) have been misused and others (such as “ectrodactyly”) have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Transverse limb deficiency (reduction) may be seen in association with amniotic bands. When both are present, both conditions should be coded.

Rudimentary or nubbin toes may be present at the distal end of a transverse limb deficiency (reduction). Their presence alone does not change the classification of the defect as transverse.

Joint contractures or clubfoot/clubhand are commonly seen in association with longitudinal limb deficiencies.

Intercalary deficiency (phocomelia) has been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal deficiency. Intercalary defects also may occur without exposure to thalidomide.

Limb deficiency is one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Oromandibular-Limb Hypogenesis spectrum, which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), and sixth and seventh cranial nerve palsies (Moebius sequence).