بسم الله الرحمن الرحيم

Radiographic Evaluation of Orodental and Craniofacial Anomalies Associated with Limb Reduction Defects

THESIS
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LIST OF ABBREVIATIONS

AER	: Apical Ectodermal Ridge
Ag	: Antigonion
ANS	: Anterior nasal spine
Ar	: Articular
BMP	: Bone Morphogenic Protein
CBC	: Complete Blood Count
CCD	: Charged couple device
Ceph	: Cepalometric Radiography
Cephs	: Cephalograms
Cd	: Condylon
DR	: Duane radial syndrome
Ds	: Disorganization syndrome
del	: Deletion
Eu	: Euryon
EUROCAT	: European Surveillance of Congenital Anomalies
Fgf	: Fibroblast growth factor
FHUF	: Femoral hypoplasia with Unusual Facies
Gn	: Gnathion
Go	: Gonion
ICBD	: International Clearing House of Birth Defects
ICD	: International Classification of Diseases
IW	: Interorbital Width
J	: Jugale
K.V	: Kilovolt
Lo	: Latero orbitale
LRDs	: Limb Reduction Defects
M.A	: Milliampere
Me	: Menton
MMA	: Maxillary Mandibular Plane Angle
MMT	: Mandibular Maxillary Transverse relation
Mo	: Medio orbital
MP	: Mandibular Plane
N	: Nasion
NBDPS	: National Birth Defects Research & Prevention Studies
NC	: Nasal Cavity
NW	: Nasal Width
OMIM	: Online Mendelian Inhertance in Man
OW	: Orbital
PNS	: Posterior Nasal Spine
Pog	: Pognion
PP	: Palatal Plane
PSP	: Photostimulable phosphor plate

S	: Sella
Shh	: Sonic hedgehog
TAR	: Thrombocytopenia with Absent Radius
TMJ	: Tempromandibular Joint
TTV	: Terminal Transverse Defect
V	: Vertex
Wt	: Weight
ZPA	: Zone of Polarizing Activity

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1-Introduction

Limb reduction defects (LRDs) which are also known as limb deficiencies or absence deformities are defined as absence or severe hypoplasia of the skeletal structures of the limb **(Temtamy and McKusick, 1978)**. LRDs are one of many congenital limb malformations but they have a higher profile among the public. In the 1960's the drug Thalidomide which was prescribed then as an antiemetic drug during pregnancy caused a large number of LRDs cases which **McCredie (2009)** refere to as the "thalidomide catastrophe". Again in the early 1990s the prenatal test chorionic villus sampling done in the early weeks of pregnancy was blamed for causing LRDs **(Evans and Wapner, 2005)**.

LRDs are also common; they have an estimated prevalence rate that ranges from 1 in 500 to 1 in 1000 human live births **(Wilkie, 2003)**. Two thirds of LRDs are considered disabling **(Vuylsteek and Hallen, 1994)**.

Craniofacial and orodental abnormalities are included in the diagnostic criteria of some LRDs and with the expanding contribution of radiology in diagnosis, a look at the craniofacial and orodental roentogenic manifestations were due necessary to help extend LRDs' comprehensive examination and cover previously uninvestigated aspects of this developemental disabling chronic problem.

2- Review of Literature

2-1. Skeletal anatomy of the limbs and patterns of limb reduction defects

Skeletal anatomy of the limbs **(Gray, 2000; Hartwig, 2008; Tank, 2009)**

The upper limb consists of four parts (Fig. 1): the shoulder, the arm, the forearm and the hand. The scapula forms the shoulder. It communicates also with the clavicle anteriorly.

The arm consists of a single long bone called the **humerus** which is the largest bone of the upper limb. It consists of a body and two extremities. The upper extremity consists of a head, a neck and two tubercles. The head is a smooth hemisphere that articulates with the glenoid fossa of the scapula to form the shoulder joint. The lower extremity of the humerus has two articular surfaces. On assuming the anatomical position (i.e. palm facing upwards) it articulates with **the radius laterally and with the ulna medially**.

The **ulna** and the **radius** (Fig.1) constitute the forearm. The ulna appears longer yet weaker than the radius. Its upper extremity is larger than the lower. It is firmly attached to the humerus to form the elbow joint. The radius's lower extremity is more prominent than the upper as it contributes to the movement of the wrist and hand.

The **hand** (Fig.2) consists of three parts; the wrist (the carpus**)**, the palm (the metacarpus) and the fingers. They constitute 27 bones. The **carpus** consists of eight small bones arranged in two rows, four bones each. The **metacarpus** is formed of five metacarpal bones numbered from I to V. The metacarpal bases articulate with the wrist bones, while

the heads articulate with the fingers and form the knuckles that appear when the fist is clenched. The fingers consist of 14 phalanges.

The **phalanges** are arranged in three rows per finger except the thumb. The first row (metacarpal related) is termed the proximal row, next is the middle and finally the distal row. The digits have terms (thumb, index, middle, ring, little) but are also numbered from I to V starting from the thumb.

The lower limb (Fig.1) consists of four parts: the hip, the thigh, the leg and the foot. The thigh consists of a single large bone called the **femur** which is the longest, strongest and largest bone of the whole body. It is made up of a body and two extremities. The head communicates with the pelvis to form the hip joint. The lower extremity is larger than the upper as it will form the knee joint. The patella is a large triangular bone which is responsible for knee joint protection.

The leg consists of the **tibia** and the **fibula** (Fig.1)**. The tibia is present medially**. It is the second longest bone of the body. **The fibula is found laterally**. It is the slenderest of all long bones. It is connected to the tibia from both its ends. It doesn't participate in the knee joint but forms the lateral part of the ankle (tarsus). The **foot** (Fig.3) consists of the tarsi which are of seven bones not eight. Starting medially the metatarsals are numbered I to V and the 14 phalanges are arranged in three rows; the proximal, middle and distal with the exception of the big toe (hallus) which is made up of only two rows.

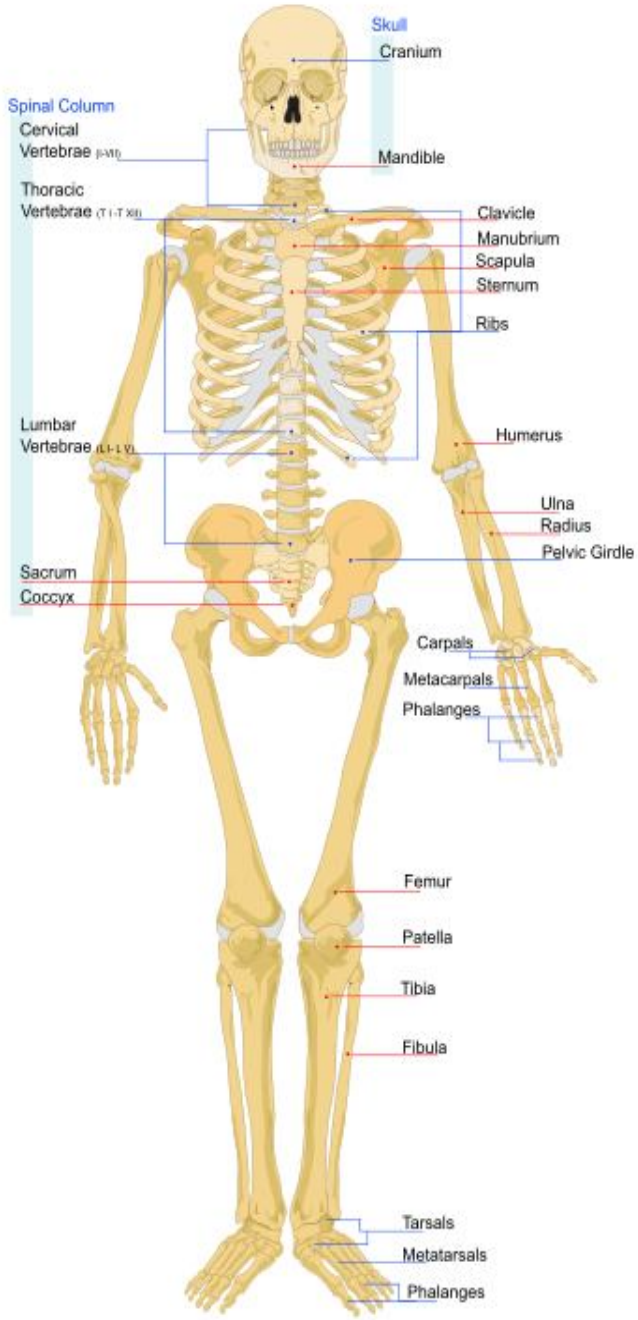

Fig.1 The human skeleton

wiki.chainofthoughts.com/dt/en/Human%20skeleton