

Contents

Introduction	1
1 Development of the Normal Fetal Skeleton	
Introduction	3
2 Differential Diagnosis of Single Skeletal Defects	
Introduction	31
Limbs	
Amelia – Amputation – Phocomelia	32
Radius and/or Thumb: Aplasia, Hypoplasia	36
Radio-ulnar Synostosis	40
Ulna: Aplasia, Hypoplasia	42
Humerus: Aplasia, Hypoplasia	44
Tibia: Aplasia, Hypoplasia	46
Fibula: Aplasia, Hypoplasia	48
Femur: Aplasia, Hypoplasia	50
Femur: Bowing	53
Stippled Epiphyses – Stippled Ossification of Cartilage	56
Hands/Feet: Absent	58
Hands/Feet: Split/Cleft/Ectrodactyly	60
Hands/Feet: Preaxial Polydactyly	63
Hands/Feet: Postaxial Polydactyly	66
Skull	
Premature Cranial Synostosis/Cloverleaf Skull	70
Unossified and Hypo-ossified Calvaria	73
Encephalocele	75
Anencephaly/Myelomeningocele/Spina bifida	78
Spine	
Vertebral Segmentation Defects/Hemivertebrae/Vertebral Fusion	80
Pelvic-Sacral Abnormalities	
Sacral Agenesis/Hypogenesis/Caudal Dysplasia; Pubic Bone Dysgenesis	86
Coronal Clefts of Vertebral Bodies	90
Platyspondyly	93
Ventral Body Wall Defects	
Ectopia Cordis	94
Ventral Wall Defects/Omphalocele/Gastroschisis	96
3 Skeletal Dysplasias	
Introduction	99
A. Skeletal Dysplasias with Prominent Shortness of the Tubular Bones	
Thanatophoric Dysplasia I	100
Thanatophoric Dysplasia II	102
Achondroplasia	104
Metatropic Dysplasia	106
Fibrochondrogenesis	108
Schneckenbecken Dysplasia	110
Lethal Metaphyseal Chondrodysplasia, Sedaghatian Type	111
Achondrogenesis 1A	112
Achondrogenesis 1B	113
Achondrogenesis Type 2/Hypochondrogenesis	114
Platyspondylic Dysplasia, Torrance-Luton Type	116
Kniest Dysplasia/Spondyloepiphyseal Dysplasia Congenita	118

B. Skeletal Dysplasias with Congenital Bowing and Normal Bone Density	
Dyssegmental Dysplasia	120
Schwartz-Jampel Syndrome	122
Cartilage-Hair Hypoplasia	123
Campomelic Dysplasia	124
C. Skeletal Dysplasia with Decreased Bone Density, with or without Bowing	
Antley-Bixler Syndrome	126
Osteogenesis Imperfecta	128
Infantile Hypophosphatasia	131
Jansen Metaphyseal Dysplasia	133
Stüve-Wiedemann Syndrome	134
Oto-palato-digital Syndrome Type II	136
D. Skeletal Dysplasias with Slender Tubular Bones	
Fetal a/Hypokinesia Sequence	138
Osteocraniostenosis	140
Gracile Bone Dysplasia	142
Microcephalic Osteodysplastic Primordial Dwarfism, Type 1	143
Microcephalic Osteodysplastic Primordial Dwarfism, Type 2	144
E. Skeletal Dysplasias with Thick Tubular Bones	
Mucopolidosis II	145
Infantile Cortical Hyperostosis	146
F. Skeletal Dysplasias with Short Ribs, with or without Polydactyly	
SRP, Saldino-Noonan/Verma-Naumoff Type	147
SRP, Beemer-Langer Type	149
Asphyxiating Thoracic Dysplasia	150
SRP, Majewski Type	152
Ellis-van Creveld Syndrome	153
G. Chondrodysplasia Punctata Group	
Greenberg Dysplasia	155
Chondrodysplasia Punctata, X-linked Dominant Type	156
Chondrodysplasia Punctata, X-linked Recessive Type	158
Chondrodysplasia Punctata, Rhizomelic Type	159
H. Skeletal Dysplasias with Multiple Dislocations	
Larsen Syndrome	161
Atelosteogenesis	163
Desbuquois Dysplasia	165
I. Skeletal Dysplasias with Prominent Involvement of the Humeri or Clavicles	
De la Chapelle Dysplasia	167
Diastrophic Dysplasia	168
Omodysplasia	170
Cleidocranial Dysplasia	171
K. Dysplasias with Prominent Involvement of the Mesial and Acral Skeleton	
Mesomelic Dysplasia, Langer Type	173
Robinow Syndrome	174
Nievergelt Syndrome	176
Grebe Dysplasia	177
L. Skeletal Dysplasias with Increased Bone Density	
Infantile Osteopetrosis	179
Blomstrand Syndrome	181
Lenz-Majewski Hyperostotic Dysplasia	182
Annex	184
Subject Index	209