

1. Record Nr.	UNINA9910809212603321
Titolo	Human malformations and related anomalies / / edited by Roger E. Stevenson [and five others]
Pubbl/distr/stampa	New York, New York : , : Oxford University Press, , 2016 ©2016
ISBN	0-19-938605-6 0-19-938604-8
Edizione	[Third edition.]
Descrizione fisica	1 online resource (1001 p.)
Collana	Oxford Monographs on Medical Genetics ; ; 66
Disciplina	616.042
Soggetti	Abnormalities, Human - Prevention Abnormalities, Human - Research
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Cover; OXFORD MONOGRAPHS ON MEDICAL GENETICS; HUMAN MALFORMATIONS AND RELATED ANOMALIES; CONTENTS; PREFACE; ACKNOWLEDGMENTS; CONTRIBUTORS; INTRODUCTION; I.1. Nomenclature; I.2. Etiology and Pathogenesis; I.3. Classification and Coding; I.4. Genetic Causes of Congenital Anomalies; I.5. Environmental Causes of Congenital Anomalies; I.6. Human Anomalies with Unknown Causes; I.7. Detection, Diagnosis, Evaluation, Management; 1 LIMBS; Introduction; 1.1. Limb Deficiency; 1.1a. Absence of Limb; 1.1b. Absent Radius; 1.1c. Absent Ulna; 1.1d. Absence and Hypoplasia of the Humerus 1.1e. Absence and Hypoplasia of the Tibia1.1f. Absence and Hypoplasia of the Fibula ; 1.1g. Absence and Hypoplasia of the Femur; 1.1h. Terminal Transverse Limb Deficiency; 1.2. Synostosis; 1.2a. Carpal Coalition and Tarsal Coalition; 1.2b. Metacarpophalangeal and Metatarsophalangeal Synostosis; 1.2c. Proximal Symphalangism; 1.2d. Distal Symphalangism; 1.2e. Humeroradial Synostosis; 1.2f. Radioulnar Synostosis; 1.2g. Tibiofibular Synostosis; 1.2h. Sirenomelia; 1.3. Constriction Rings; 1.4. Excessive Partitions, Duplications, and Accessory Bones; 1.5. Bowing of Long Bones; 1.6. Short Stature 1.7. Tall Stature1.8. Limb Overgrowth; 1.9. Increased Bone Density; 1.10. Decreased Bone Density; 1.11. Osteolysis; 1.12. Anomalies of the

Patella; 1.13. Arthrogryposis; 1.13a. Amyoplasia; 1.13b. Distal Arthrogryposis; 1.13c. Fetal Akinesia Phenotype; 1.13d. Pterygium; 2 | HANDS AND FEET; Introduction; 2.1. Polydactyly; 2.1a. Preaxial Polydactyly; 2.1b. Postaxial Polydactyly; 2.1c. Mesoaxial Polydactyly; 2.2. Syndactyly; 2.2a. Syndactyly Type I; 2.2b. Syndactyly Type II; 2.2c. Syndactyly Type III; 2.2d. Syndactyly Type IV; 2.2e. Syndactyly Type V; 2.2f. Complete Syndactyly
2.2g. Cenani-Lenz Syndactyly2.2h. Symbrachydactyly; 2.3. Brachydactyly; 2.3a. Brachydactyly Type A1; 2.3b. Brachydactyly Type A2; 2.3c. Brachydactyly Type A3; 2.3d. Brachydactyly Type B; 2.3e. Brachydactyly Type C; 2.3f. Brachydactyly Type D; 2.3g. Brachydactyly Type E; 2.4. Osseous Deficiencies of the Hands and Feet; 2.4a. Preaxial Deficiency; 2.4b. Postaxial Deficiency; 2.5. Terminal Transverse Deficiency; 2.6. Split-Hand/Foot Malformation; 2.7. Macrodactyly; 2.8. Camptodactyly; 2.9. Clubfoot; 3 | PECTORAL AND PELVIC GIRDLES; Introduction; 3.1. Clavicular Hypoplasia or Aplasia
3.2. Clavicular Pseudoarthrosis3.3. Altered Shape and Other Abnormalities of the Clavicle; 3.4. Sprengel Anomaly; 3.5. Glenoid Hypoplasia; 3.6. Anomalies of the Pelvic Bones; 3.7. Developmental Dysplasia of the Hip; 3.8. Coxa Varus; 3.9. Coxa Valga; 4 | SPINE AND THORACIC CAGE; Introduction; 4.1. Occipitalization of the Atlas; 4.2. Aplasia/Hypoplasia of the Odontoid Process of the Axis; 4.3. Klippel-Feil Anomaly; 4.4. Segmentation Defects of the Vertebrae; 4.5. Altered Vertebral Body Contour; 4.6. Sagittal Clefts of the Vertebrae; 4.7. Coronal Clefts of the Vertebrae
4.8. Spondylolysis and Spondylolisthesis

Sommario/riassunto

The central theme of this text is to provide information on individual anomalies & to connect these anomalies to the malformation syndromes & associated problems, primarily through the use of differential diagnostic tables.
