Hum Genet (2015) 134:1271
DOI 10.1007/s00439-015-1597-7

BOOK REVIEW

Ghazi M Rayan and Joseph Upton III: Congenital Hand Anomalies and Associated Syndromes

Springer, 2014, hardcover, 500 pages, ISBN 978-3-642-54610-5

Ruth Newbury-Ecob[1]

Published online: 4 September 2015

This is a long-awaited comprehensive review of congenital hand anomalies and associated syndromes that will appeal to clinical geneticists and hand surgeons alike. The illustrations are outstanding, with a combination of photographs, X-rays, illustrative diagrams, plaster moulds and CT scans. As this was written by two hand surgeons, the clinical genetics reader may find some of the surgical photographs unsettling, but they are helpfully linked to pre-operative photographs and serve a purpose reminding us of the internal anatomy of the limb and how this, too, may be abnormal. There are descriptions of 127 associated syndromes and 37 congenital differences. Although this is a highly specialised field, the breadth of conditions covered alludes to the high frequency of limb anomalies in genetic syndromes. Syndromic descriptions include photographs of the syndromologist, hallmarks, background, aetiology and presentation, and a list of associated anomalies.

A clear systematic approach is used with Chapters 1 subdivided into tumours, enchondromas, osteochondromas, neurofibromas, vascular malformations, congenital contractures, and congenital joint laxity/instability. Chapters 2 and onwards are classified by the anatomical location (elbow, forearm, hand, thumb, digits, and skin).

The book starts with a reminder from the 1982 Lamb study, of a birth incidence for upper limb malformations of 11 per 10,000 live births, 51 % being bilateral and 17 % associated with multiple malformations. Limb defects are more prevalent in boys, with pre-term, post-term and multiple births, and in the offspring of older mothers.

Classifications are based on those of the American Society for Surgery of the Hand and the International Federation of Societies for Surgery of the Hand. The classification encompasses several categories, including failure of formation, defective differentiation, duplication, undergrowth and overgrowth, constriction bands and general skeletal anomalies. This classification blends well with our current understanding of the aetiology of limb defects. The book covers aetiology only minimally but briefly alludes to the key molecular mechanisms. Given the rapid pace of progress in understanding the molecular genetics of limb development, it would be difficult for a text book to remain in date and therefore appropriate for the reader to look elsewhere.

I would strongly recommend this as an educational resource to understand the use of specific terms describing hand malformations and as a practical tool to aid with diagnosis of hand anomalies. The resource of photographs is unique.

✉ Ruth Newbury-Ecob
Ruth.Newbury-Ecob@UHBristol.nhs.uk

[1] Department of Clinical Genetics, University Hospitals Bristol NHS Foundation Trust, St Michael's Hospital, Southwell St, Bristol BS2 8EG, UK