mild case' by Turolla *et al.*[1] The authors reported a 6 month old male child with a combination of craniofacial dysmorphism, postaxial polydactyly of the toes, and agenesis (or hypoplasia) of the corpus callosum. Preaxial polydactyly and other less frequent anomalies present in the acrocallosal syndrome (ACS) were absent. According to the authors, this case was the fifth patient with mild expression of ACS and they stressed the importance of the identification of patients with a less severe clinical picture of ACS.

During the past three years we have had the opportunity to examine in Leuven two other unrelated male patients with mild expression of ACS.

The first boy was the first child of healthy, unrelated parents. Pregnancy was normal but delivery was induced at 34 weeks after the echographic diagnosis of macrocephaly and a midline porencephalic cyst. Birth weight, length, and head circumference were 3520 g, 53·5 cm, and 38·4 cm respectively. The forehead was broad and bulging, the anterior fontanelle was large, and the nasal bridge was broad and flat. There was preaxial polysyndactyly on the right foot with complete duplication of the hallux and syndactyly of the first and second toes, a varus deformity of the right foot, and lateral bowing of the right tibia. Clinical and radiographic examinations of the left foot and both hands were normal. Nuclear magnetic resonance imaging showed complete agenesis of the corpus callosum and a large interhemispheric cyst. Chromosomal examinations on a peripheral blood lymphocyte culture showed a normal 46,XY male karyotype after G banding. No major problems occurred in the following months. At the age of 2 years 8 months weight was 15·6 kg, length 91·5 cm, and head circumference 54·2 cm (2 cm above the 97th centile). Psychological evaluation showed an intellectual level of 32 months at the chronological age of 34 months (IQ 94, Stutsman scale). Three months ago a normal brother was born.

The history of the second boy is identical. He was the first child of healthy, unrelated parents. Prenatal ultrasound showed intracranial cysts and he was born by elective caesarean section at 39 weeks. At the age of 7 weeks he was admitted to the paediatric department for further evaluation. He was macrocephalic with a high, broad forehead and a large anterior fontanelle (weight 4·5 kg, length 55 cm, and head circumference 41·3 cm). There was preaxial polysyndactyly of the right foot with hallux duplication and syndactyly of the first, second, and third toes. NMR imaging showed agenesis of the corpus callosum and the presence of one large and several small interhemispheric cysts. Now, at the age of 13 months, weight and length are on the 75th to 90th centiles and head circumference is 53 cm (3 cm above the 97th centile). Denver developmental scale showed normal psychomotor development for his age.

The present two unrelated male patients are other (identical) examples of minor expression of ACS: they combine the craniofacial features with preaxial poly(syn)dactyly limited to the right foot and complete agenesis of the corpus callosum with large interhemispheric cyst(s). Moreover, the developmental level in both boys was within normal limits. This indicates also that the variability in long term mental performance in the ACS is greater than has been documented up to now.

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1 Turolla L, Clementi M, Tenconi R. How wide is the clinical spectrum of the acrocallosal syndrome? Report of a mild case. *J Med Genet* 1990;27:516–8.

BOOK REVIEWS

Advances in Human Genetics. Volume 19. Ed H Harris, K Hirschhorn. (Pp 339; $65·00.) New York: Plenum Press. 1990.

It is sobering to reflect on the changes that have occurred in human and medical genetics during the two decades which have elapsed since this series was first launched. Although more clinically orientated than many of its predecessors, this 19th volume will be of equal interest to clinician and scientist alike. The original editors are still in harness and can be just as proud of this latest offering as of the previous 18 volumes. They have successfully commissioned five authoritative monographs, each of which provides a comprehensive contemporary review of an area of direct clinical relevance. Much of the information is not readily available elsewhere and it is particularly valuable to have such diverse yet topical material gathered together in a single source.

The opening chapter alone justifies the not unreasonable purchase price. Drs Spranger and Maroteaux have been persuaded to share their vast experience and unique collection of lethal dwarfing syndromes. The result is a definitive review and classification of the lethal chondrodysplasias running to more than 100 pages with over 150 radiographic illustrations. Appropriately this is followed by a refreshingly comprehensible discussion of the nature of collagen and the spectrum of mutations observed in osteogenesis imperfecta. The remaining three chapters provide in depth accounts of inherited platelet disorders, immunoglobulin A deficiency, and, finally, a bewildering array of conditions associated with defective mitochondrial metabolism.

Keeping abreast of burgeoning publications on human and medical genetics is rapidly becoming an impossible task. However, with volumes such as this, help is at hand. This compilation of well written and exhaustive reviews will make a thoroughly worthwhile addition to any personal or departmental library.

I D YOUNG

Human Achondroplasia—A Multidisciplinary Approach. Ed Nicoletti, Kopits, Ascani, McKusick. (Pp 491; $95·00.) New York: Plenum Press. 1988.

This book contains the papers given at the First International Symposium on Human Achondroplasia held in Rome in November 1986. The approach is multidisciplinary, covering genetics, histopathology, clinical features, natural history, and the problems of anaesthesia. The greater part of the book

reports surgical complications and management with a section on spinal disorders followed by the techniques and results of leg lengthening. The final papers deal with social and psychological aspects of the disorder.

The first third of the book and the final papers on social and psychological aspects are principally of interest to paediatricians, clinical geneticists, and basic scientists interested in the skeletal dysplasias, and there were many distinguished speakers at this conference. However, there are papers dealing with the principles rather than details of surgery which make interesting and useful reading for the non-surgical specialist. (These 'details' are now four years out of date and would need to be read today with this fact in mind.)

It is uncommon to find this multidisciplinary approach and the book should be on the shelves of clinical genetic libraries as an example of what can be achieved. Many of the problems discussed here are relevant also to other types of skeletal dysplasia where short stature is a major feature.

The book is neatly produced, with illustrations of which some are excellent and nearly all adequate for their purpose.

RUTH WYNNE-DAVIES

Epidermolysis Bullosa: A Comprehensive Review of Classification, Management and Laboratory Studies. Eds G C Priestley, M J Tidman, J B Weiss, R A J Eady. (Pp 198; £20·00.) Crowthorne, England: Dystrophic Epidermolysis Bullosa Research Association. 1990.

This paperback contains the proceedings of a meeting held in January 1989. There are 43 chapters and 53 named contributors, providing a very wide review of many different scientific and clinical aspects of epidermolysis bullosa. The book is clearly printed with scattered black and white illustrations.

The main difficulty with using and reviewing this book is that attempts by the editors to impose some logic on the order of the chapters is frequently sabotaged by subjects suddenly appearing at inappropriate times. Reviews are muddled in with clinical reports and different aspects of counselling are scattered throughout the book. If you persevere, however, you will be rewarded by several excellent scientific reviews of the state of laboratory research and many practically helpful guides to different aspects of clinical management.

All involved in the medical and nursing care of patients with epidermolysis bullosa may benefit from the specific advice given about the management of dental problems, dysphagia, and constipation. The importance of adequate nutrition is stressed. Special problems of anaesthesia, physiotherapy, and management of eye problems provide useful practical tips. The review by Tidman on the occurrence of malignancy in epidermolysis bullosa gives an important message, reminding doctors and others caring for these patients to be aware of this potential complication. I particularly enjoyed Atherton's thoughtful account of the meaning and medical responsibilities of counselling and his reviews on neonatal management and the lack of progress in effective systemic treatment.

The case reports published are of variable quality. Some are very short, but enlivened by the discussion transcripts. There are some excellent scientific reviews, in particular a closely referenced article by Fine on the significance of basic membrane related antigens in epidermolysis bullosa and that by Bauer *et al* on the role of proteinases. The two chapters by Paavonen *et al* on the measurement of creatine kinase and lactate dehydrogenase could have benefited from a more detailed discussion of the importance of this area in the understanding of epidermolysis bullosa. The final chapter is the proceedings of a session entitled 'Recent advances and future opportunities', providing a compact but useful 'state of the art' summary.

Chapters on the classification of epidermolysis bullosa, counselling, prenatal diagnosis, and the role of the geneticist in epidermolysis bullosa provide information of specific relevance to the clinical geneticist.

DEBRA should be congratulated on organising the conference on which this book is based and the editors for making the information given at the conference widely available.

ANDREW Y FINLAY

NOTICE

The 8th International Congress of Human Genetics

This Congress, sponsored by the American Society of Human Genetics, will be held on 6 to 11 October 1991 at the Washington Convention Center in Washington, DC. The deadline for receipt of abstracts is 1 April 1991. For abstract and registration forms or additional information, contact: M Ryan, Meetings Manager, ICHG, 9650 Rockville Pike, Bethesda, MD 20814, USA. (Tel (301) 571–1825; Fax (301) 530–7079).