mild case’ by Turolla et al. 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 porencephaly 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 5 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, Stanford 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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BOOK REVIEWS


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


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