Topics in Pediatric Genetic Pathology. The Enid Gilbert-Barness Festschrift

Topics in Pediatric Genetic Pathology is a compilation of 45 papers put together as a Festschrift for Enid Gilbert-Barness. They have also been published as a supplement to the American Journal of Medical Genetics.

As its title suggests, the main thrust of this book is the description and discussion of visceral abnormalities as manifestation of congenital or inherited disorders. Accounts of genetic disorders are all too often spoiled when careful external description accompanied by high quality radiographs and painstaking biochemical investigations are published with brief, vague descriptions of visceral abnormalities and usually no account of histological appearances. This book goes a long way to redress this problem. It presents useful accounts of the range of necropsy findings in some well recognised conditions as well as detailed descriptions of pathological abnormalities in individual cases of less common disorders. There is a separate section comprising four papers on skeletal dysplasias which describe histological findings as well as clinical and radiographical abnormalities and assess the contribution of all three to differential diagnosis within each group of disorders.

The short section on biochemical and metabolic aspects of developmental pathology provides an assessment of the appropriate use of potentially expensive investigations. The papers on cardiac morphogenesis reviews and introduces concepts which have more general application to pathological development.

The first section, which covers general principles and methods, contains some papers which are highly speculative and others which, by their very nature, will become rapidly outdated. The papers covering DiGeorge anomaly and posterior cervical hygroma are, however, of both immediate and probably lasting usefulness.

This book is attractively presented, well illustrated, and carefully edited to provide easy reading without obliterating individual style. This book has much to offer the clinical geneticist and those, whether pathologist or geneticist, called upon to examine the dysmorphic fetus. It should be particularly useful to both geneticist and pathologist in the planning and assessment of necropsy examination of the malformed stillbirth or infant.

Alan E H Emery

The Mammalian Y Chromosome: Molecular Search for the Sex-Determining Factor

What a strange chromosome the human Y is! It has homology to the X and to autosomes, of both man and other species, but scarcely any homology to the Y of other species. Its only certain function is to carry the testis determining gene (TDF), but now that this has been cloned it turns out to have a counterpart on the X. The only other adequately characterised gene on the Y, MIC2, shows autosomal inheritance, being located in the X-Y pairing region at the tip of the short arm. This region and the adjacent TDF, which look like the functional core of the Y, might be expected to be conserved between species, but in the chimpanzee the pairing region is at the other end of the chromosome. The rest of the human Y seems to have been cobbled together over recent evolutionary time from spare bits of the X and autosomes.

This book contains the papers and posters presented at the meeting of the British Society for Developmental Biology at Oxford in March 1987, and it touches on all the puzzles of the mammalian Y chromosome. The mystery of XX males and XY females has largely evaporated; in most cases maleness goes with the presence of region 1 of the short arm. Several papers (Ferguson-Smith, Muller, P N Goodfellow, Weissenbach) describe the ordering of Y probes by deletion mapping and their use to demonstrate the abnormal X-Y interchanges which are responsible for most XX males and XY females. XX true hermaphrodites and some familial XX males remain unexplained. The cloning of TDF by Page, of course, came too late for inclusion, but the background is well explored.

The other genes of the Y chromosome are less well characterised. The H-Y gene maps near Tdy (=TDF) in the mouse, but in the long arm in man; in each case it coincides with a gene necessary for spermatogenesis. P J Goodfellow describes the evidence for a pseudoautosomal XGR locus regulating the level of both Xg and MIC2 expression. There is at least part of a steroid sulphatase (STS)