The book is divided into three sections. In 200 pages and 100 illustrations the first section covers the fundamentals of genetics including cell structure, molecular genetics, prokaryotic and eukaryotic cells, mitochondria and the mitochondrial genome, linkage analysis and inheritance, chromosomes, and gene expression. The second section addresses the more medical aspects of genetics with some excellent summaries of diseases in which the disease is described in text and a series of diagrams illustrates the clinical picture (with a photograph) and the molecular basis of the condition. For example, the pages covering neurofibromatosis 1 and 2 have a page of text describing the main features of each condition. The accompanying diagrams show a list of facts about NF1, three clinical photographs and three diagrams of the NF1 gene, the NF1 gene product, and a regional map of the NF2 region. The NF2 gene is not described, presumably because this was identified too recently to be included in the book. The third section of the book describes genomic organisation in prokaryotes and eukaryotes. There are comparative chromosome maps of human, mouse, cat, and cow, and human chromosome disease maps along with a list of diseases and map locations in alphabetical order.

This whole book has around 400 pages and will fit into a pocket. In that restricted space the whole of genetics is covered in remarkable detail and in a very readable format. It was originally written in German and there are occasional words which have not been translated, but this does not detract from the quality of this book. This is not just another book about genetics but is an excellent addition to the available texts.

DOROTHY TRUMP

Color Atlas of Congenital Malformation Syndromes. Michael Baraitser, Robin M Winter. (Pp 233; £65.00.) UK: Mosby-Wolfe. 1996. ISBN 0-7234-2073-4.

Baraitser and Winter have translated some of their extensive experience in diagnostic dysmorphology into this excellent book. It is written in an atlas type format, with over 400 malformation syndromes being described. Each entry contains a succinct description of the syndrome, including a few key references,

and concentrating on areas of clinical and genetic relevance. Although these "thumb nail" descriptions are useful, the real strength of this book lies in the 900 illustrative colour photographs which accompany them. Clinicians grappling with the problems of making a diagnosis in dysmorphic children will find these photographs invaluable. They will also be helped by the problem oriented approach of the book; syndromes are grouped into chapters based on their principal mode of presentation.

Who should buy this book? In their introductory chapter, Baraitser and Winter state that the aim of the book is to improve the ability of the reader to make "gestalt" diagnoses. The high quality of the book is such that the simplest answer to this question is, perhaps, anyone who wants to improve this skill. It will certainly be a useful addition to the bookshelves of most clinical geneticists and many paediatricians.

EVAN REID

NOTICE

Human gene mutation database

The *Human gene mutation database (HGMD)* (supported by SmithKline Beecham) is now available on the worldwide web at <http://www.cf.ac.uk/uwcm/mg/hgmd0.html>.

HGMD represents an initial attempt to collate the majority of known (published) human germline gene mutations. HGMD thus comprises single base pair substitutions and short (<20 bp) deletions in human nuclear genes causing inherited disease. Somatic gene mutations and mitochondrial genome mutations are not included. Each mutation is entered only once to avoid confusion between recurrent and identical by descent lesions.

Data acquisition has been accomplished by a combination of manual and computerised search procedures. In excess of 250 journals are scanned on a weekly/monthly basis for articles describing germline mutations. Coverage is limited to original published reports although some data are taken from "Mutation

Updates" and review articles. Mutations reported in abstract form are not generally included. All entries comprise the disease state as specified in the original article, the gene name, symbol (as used by the *Genome database*) and chromosomal location, and a reference to the first published report of the mutation.

HGMD currently (June 1996) contains >5400 different single base pair substitutions from >400 gene loci, each represented by the nucleotide and ensuing amino acid change plus the number of the affected codon. Single base pair substitutions outside coding regions (that is, in promoter regions, splice sites, introns, or untranslated regions), complex substitutions involving the replacement of more than one base at a specific site, and silent mutations within the coding region which do not alter the encoded amino acid are not yet included. (Hypertext links to 18 WWW based Locus Specific Mutation Databases are however provided and serve to augment coverage of these loci.) Mutations inferred from amino acid sequencing have also been excluded since, in the absence of direct DNA analysis, some ambiguity may exist as to the DNA sequence change involved.

HGMD currently also contains over 1100 different microdeletions derived from some 220 gene loci, each represented by the deleted bases ± 10 flanking bases and their location in the respective gene sequence. Deletions over 20 bp in size and complex rearrangements involving the insertion as well as deletion of bases are not included.

Originally established for the study of mutational mechanisms in human genes,¹ HGMD has now acquired a much broader utility. For the categories of mutation covered, HGMD represents the most comprehensive available reference source to the spectrum of mutations underlying human genetic disease and provides information of importance to researchers in human molecular genetics, physicians, and genetic counselors. DAVID N COOPER and MICHAEL KRAWCZAK *Institute of Medical Genetics, University of Wales College of Medicine, Heath Park, Cardiff CF4 4XN, UK.*

1 Cooper DN, Krawczak M. *Human gene mutation*. Oxford: Bios Scientific Publishers, 1993.