LETTER TO THE EDITOR

Hypohidrotic ectodermal dysplasia with hypothyroidism

In the August 1989 edition of the Journal, Fryns *et al*[1] published a case of hypohidrotic ectodermal dysplasia with hypothyroidism, and referred to a previous case by Pabst *et al*[2] of two male sibs with a similar condition.

As the Fryns case was male, they suggested that further case reports were needed before X linkage could be excluded.

In 1986, we published a case of a girl with this condition and did suggest in that article that recessive inheritance was more likely.[3]

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1 Fryns JP, Chrzanowska K, Van Den Berghe H. Hypohidrotic ectodermal dysplasia, primary hypothyroidism, and agenesis of the corpus callosum. *J Med Genet* 1989;26:520–1.
2 Pabst HF, Groth O, McCoy EE. Hypohidrotic ectodermal dysplasia with hypothyroidism. *J Pediatr* 1981;98: 223–37.
3 Pike MG, Baraitser M, Dinwiddie R, Atherton DJ. A distinctive type of hypohidrotic ectodermal dysplasia featuring hypothyroidism. *J Pediatr* 1986;108:109–11.

BOOK REVIEWS

Genetic Engineering 7. Ed Peter W J Rigby. (Pp 127; £14.00.) New York, London: Academic Press. 1989.

The first volume in this series was published around 10 years ago, and under its two excellent editors the subsequent volumes in the series have moved with the times and kept pace with the many new developments in genetic engineering. Clinical molecular genetics has meanwhile also moved forward at a sometimes stunningly fast rate. However, the topics covered in the series, and those of most interest to medical geneticists, have somewhat diverged and this is illustrated in the three topics covered in this volume.

Chapter 1 discusses the molecular biology of the kineto plastidae, concentrating particularly on African trypanosomes. Chapter 2 deals with cloning antigen genes from malarial parasites and *Leishmania* species, but includes some very interesting introductory material on malaria and the current state of knowledge of the human receptors for malarial parasites.

The final chapter deals with the production of foreign proteins in mammalian cells, covering mammalian cell expression systems, regulation of protein production, and large scale culture and production. As with the chapter on malaria, there will be much of interest in a general way to medical geneticists, as the author works for Cell Tech Limited, and naturally some of the molecules in which they are most interested, such as tissue plasminogen activator, are of great medical interest. In summary, this is a specialised book for the genetic engineer and, of course, it is always important for the medical geneticist to know the exciting developments which will affect them in the future.

S MALCOLM

Molecular Biology of the Eye: Genes, Vision, and Ocular Disease. Ed J Piatigorsky, T Shinohara, P S Zelenka. (Pp 510; $96·00.) New York: Alan R Liss. 1988.

This volume is the outcome of a UCLA Symposium held in February 1988 which covered a wide range of topics on the molecular biology of the eye aimed principally at visual scientists and molecular biologists. The subject matter spans almost the entire range of molecular biological techniques only loosely held together by a common focus on the visual system. Many of the chapters strongly evoke the power of these techniques and make fascinating reading, apart from their interest to visual scientists. The text is generally concise and easy to read with only occasional editorial lapses associated with camera ready proofs.

The subject matter is introduced on a futuristic note with a paper on gene therapy by C T Caskey and co-workers. The current state of knowledge on phototransduction is reviewed by several authors and the power of site directed mutagenesis in elucidating structure-function relationships in rhodopsin is described by H G Khorana and co-workers. A fascinating section on the evolution of ocular genes includes one by G Wistow and colleagues on the evolution of crystallins. This illustrates how totally unexpected findings still emerge from time to time in biology. It appears that while the alpha and beta crystallins have evolved by duplication and divergence from a non-lens specific ancestor, the other major crystallins (epsilon, delta, rho, tau) are actually encoded by genes for common cellular enzymes (for example, lactic dehydrogenase, enolase) which are overexpressed in the lens. Another evolutionary strategy is suggested for the collagen gene family (G Vogeli *et al*) in which the highly repetitive motif associated with the triple helix of type I collagen is broken up by 50 introns which, it is argued, serve to stabilise the gene by reducing opportunities for deletion, hence reducing the rate of evolutionary change.

The fundamental question of how ocular tissues differentiate is examined in a section on gene expression and differentiation. Several authors review the attempts to identify key regulatory sequences and nuclear proteins associated with tissue specific expression of lens crystallins, using gel retardation assays, deletion analysis of promoter and enhancer regions, and transient assays in cultured cell systems. The introduction of crystallin promoter sequences coupled to a marker gene into transgenic mice provides a convincing in vivo demonstration of the importance of such sequences in inducing tissue specific expression (A B Chepelinski and co-workers).

Finally, the application of molecular biological techniques to human ocular disease is discussed, with chapters on uveitis, viral infections, cataract, mouse and human retinal degenerations, and retinoblastoma. The putative retinoblastoma gene is shown by B Gallie and co-workers to contain point mutations or small deletions in a