PostScript

BOOK REVIEW

Hereditary Hearing Loss and its Syndromes, 2nd edn

Edited by H V Toriello, W Reardon, R J Gorlin. Oxford University Press, 2004, £110, pp 502. ISBN 0-19-513849-X

Although formally classified as the second edition, this is essentially the third version of the groundbreaking book by Bruce Konigsmark and Bob Gorlin entitled *Genetic and Metabolic Deafness* as originally published in 1976. Subsequent recognition of the pressing need to incorporate the rapid expansion in knowledge of new syndromes prompted the appearance of the first editon of *Hereditary Hearing Loss and its Syndromes* in 1995, as a sister publication to *Syndromes of the Head and Neck*. This new edition represents the coming of age of the marriage between molecular biology and conventional clinical genetics and provides an excellent state of the art synthesis of contemporary knowledge.

A reviewer's task in making criticism of an outstanding and definitive textbook is not an easy one. In planning this new edition the editors have resisted the temptation to tinker with a successful format to the extent that the overall structure is virtually unchanged. The existing chapter on endocrine and metabolic disorders has been split into two and the miscellaneous chapter has disappeared, to be replaced by a chapter on cardiac syndromes. Otherwise the chapter headings are as in the previous edition with much of their text reproduced, albeit with expanded sections on "heredity" to embrace the many new discoveries of the last few years. Thus the contents can be subdivided into four introductory chapters which set the scene, followed by 12 chapters describing system associated hearing loss syndromes. In general these are excellent, with each providing detailed accounts of an exhaustive list of common and rare conditions in which hearing loss can occur. All these are lavishly illustrated with ample references for those who wish to delve further.

Against this background of general excellence any possible hint of criticism might well be viewed as petty and inappropriate, so it is hoped that the editors will forgive a few personal comments. Most readers will be very familiar with the basic principles of human genetics so that on turning to the chapter on genetic counselling it was disappointing to find that this is largely limited to an explanation of traditional patterns of inheritance. The real challenge facing most clinical geneticists and genetic counsellors is how to counsel the hearing parents of a child with isolated non-syndromal hearing loss. Chapter 2 provides useful suggestions for investigation but the subsequent chapter on genetic counselling provides little in the way of assistance. True, there is a useful table (of unstated source) providing empirical risks, but with little in the way of guidance as to how these should be applied. Should these risks be modified on the basis of age of onset, laterality, asymmetry, progression, vestibular involvement, audiology or a normal Connexin 26/30 mutation analysis? Presumably they should, but how? The editors and chapter authors embrace most of the world's experts on genetic hearing loss and it is a little unfortunate that they could not expand on this crucial component of the counselling process. An overview of how genes and their products interact to facilitate the hearing process would also be useful, as would expansion of some of the sections on molecular pathogenesis in the system orientated chapters. Finally, the era when medical books can include full frontal nude photographs of children and adults must be coming to a close and one wonders how many of the stark naked adults appearing in some of the syndrome chapters gave informed consent for their publication in perpetuity.

Clearly these are minor criticisms of an excellent textbook which will provide an invaluable resource and be consulted widely. It is difficult to see how any department encountering patients with hearing loss could possibly manage without it.

I Young

CORRECTIONS

doi: 10.1136/jmg.2005.18333corr1

An error has been detected in the online mutation report by Burdon *et al* (*J Med Genet* 2004;**41**:e106). The mutation is identified in the manuscript as 226G>A in regards to the Genbank reference NM_021954. However, it should be 227G>A. The amino acid designation, R76H, is correct and this numbering error does not change any of the other results or conclusions of the article. The author apologises for this error.

doi: 10.1136/jmg.2004.013151corr1

Several errors have been detected in the electronic letter by Toyama *et al* (*J Med Genet* 2004;**41**:e74).

First, the abbreviations for Table 1 should read:

Ex, exon; (FAM)-,FAM-labelled; (HEX)-, HEX-labelled; (ROX)-,ROX-labelled; (NED)-, NED-labelled; UP, upstream; Pro, prometer; Int, intron; Fl, flanking; STR, short tandem repeat.

Second, the parenthesised section of the last sentence of the Results should read:

(7.3 ± 1.3 mmol/l (K287I) and 7.63 ± 1.0 mmol/l (M310I) compared to that of the wild type (3.8 ± 0.4 mmol/l).

In addition, in Ex4 of Table 3 the "Type" should read C235 (R79W), in Figure 1 "Euro. Am" is the abbreviation for "European American," and in Table 4 the title should read "Catalytic activity of recombinant AMPD1 expressed in *E. coli*".

We apologise for these errors.

doi: 10.1136/jmg.2004.019190corr1

The authors for the paper titled Positive association of the DIO2 (deiodianase type 2) gene with mental retardation in the iodine-deficient areas of china (*J Med Genet* 2004:**41**;585–590) have identified an error within their abstract. The second line from the results section should read: Particularly with rs255012, CC genotype frequency was significantly higher in MR cases than in controls (chi squared = 9.18, p = 0.00246). The author apologises for this mistake.