Book Reviews

Am. J. Hum. Genet. 47:595–596, 1990

Turner Syndrome. Edited by Ron G. Rosenfeld and Melvin M. Grumbach. New York: Marcel Dekker, 1990. Pp. 524. $150.00.

On the 50th anniversary of the initial report by Henry Turner, the First International Turner Syndrome Symposium was held in San Francisco. From that symposium this well-organized book was written, summarizing the present knowledge of Turner syndrome (TS). This is in fact one of the first books covering all aspects of TS that has been written.

This first symposium volume starts with Turner's original description from November 1938, which is, except for the cardiac abnormalities, an almost complete presentation of the clinical features of the syndrome. The rest of the book is divided into five sections.

Part I, "Genetics, Organogenesis and Incidence," is in seven chapters, discussing the mechanisms resulting in growth retardation and sexual underdevelopment in TS. The question is, Which genes are responsible for the phenotypical abnormalities of TS. Genes within the pseudoautosomal region of the X chromosome would be expected to behave as autosomal genes, since they escape inactivation. Some, but not all, of the features of TS are probably due to gene dosage effects from genes within the pseudoautosomal region. Two genes have been mapped within the pseudoautosomal region: the *MIC2* and the steroid sulphatase genes. Neither of these genes seems to be responsible for the TS phenotype. The critical regions for maintenance of ovarian function appear to be Xq13, Xq21, and Xq26. The critical area involved in stature seems to be the proximal Xq region, but the data are still limited. The seven chapters in this section are all very well written. Although no genes have been found to be responsible for the TS phenotype, it seems reasonable to assume that within the next 5 years those genes will be found, and probably it will be shown that TS is a gene dosage disease, similar to autosomal chromosome aberrations.

Part II, "Natural History and Associated Abnormalities," reviews in nine chapters many different aspects of TS, such as the abnormal karyotypes, the prenatal mortality, prenatal diagnosis, the possibility of egg donation, the development of ovaries, the growth and absence/prevalence of spontaneous puberty, the pathogenetic background of some TS abnormalities, the endocrine abnormalities (especially the high incidence of thyroid antibodies), and, finally, the cardiovascular involvement, especially the high frequency of aortic valve lesions. All these issues are carefully reported, and much new information is presented.

Part III is in 10 chapters, discussing the alternative causes of growth retardation that accompany the TS. The conclusion is that girls with TS are not growth hormone deficient. The skeletal abnormalities and the abnormal chondrogenesis, with development of osteoporosis, are well discussed in three chapters.

In part IV the various endocrine treatments are presented in nine chapters. The beneficial short-term effects of anabolic steroids, estrogens, and growth hormone, alone or in combination, seem to be quite clear, but the effect on the final height is still unclear. The problems of evaluating bone age in TS girls and of predicting adult final height are very well shown by J. W. Frane and B. M. Sherman. It is obvious, despite the above-mentioned problems, that there are many studies which show that girls with TS have beneficial effects from hormone therapy and should be treated with growth hormone in combination with other hormones. The most important issue concerning therapy of TS patients is whether the treatment makes them feel like successful human beings. Replacement therapy in adult women with TS seems very appropriate. The letter to Dr. Söderholm—from a woman who wrote: "Prior to therapy I felt like half a woman. The therapy has been something very positive in my life. I now feel like a total woman, who can menstruate like other age mates"—is very convincing.

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The final section presents, in six chapters, the intellectual and psychosocial development of TS women. The investigations presented show that the intellectual function of the patients with TS is within the normal range, although heterosexual behavior is delayed. The overall picture of school performance in this group is normal. In addition, they have an excellent record of obtaining and keeping employment as adults. These findings are very important for genetic counseling, especially when a TS syndrome fetus is prenatally diagnosed.

This volume provides an up-to-date survey of TS. It is much more than a report from a symposium. It is the first textbook in this field, and it is highly recommended to experts dealing with TS women. The price ($150.00) is, however, inhibiting. This is truly unfortunate, since the data presented and the quality of the presentations are excellent.

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Myotonic Dystrophy, 2d ed. By Peter S. Harper. Philadelphia: W. B. Saunders, 1989. Pp. 384. $80.00.

Professor Harper has presented a scholarly, comprehensive, well-written review of myotonic dystrophy. Myotonic dystrophy is of special importance to students of genetics because of the variability of disease expression and the potential for therapeutic intervention. Myotonic dystrophy is characterized by variable expressivity, late age at onset, and pleomorphic features. The patients can present with complaints of dysfunction involving many different organ systems. They appear in offices of a broad cross-section of medical specialities, and frequently the diagnosis is missed for generations. Although most patients present as the first case in a pedigree, examination of relatives virtually always identifies other affected individuals. This disease has been the focus of molecular genetic studies and has been assigned to the proximal long arm of chromosome 19. There are now several closely linked DNA probes for heterozygote diagnosis. Prenatal diagnosis is also possible. However, with myotonic dystrophy, the geneticist must rely very heavily on diagnoses made in relatives. Diagnosis of gene carriers is not as straightforward as it is with many other autosomal dominant inherited diseases. This complicates the linkage analysis and makes this a potentially treacherous area for nonspecialized laboratories.

Professor Harper's book provides great insight into the clinical and genetic aspects of the disease. The interweaving of clinical genetics is done superbly. Each aspect is carefully explained and referenced. Its one shortcoming is a noncritical appraisal of some of the scientific literature, relying on consensus and analogy rather than on evaluation of data presented. This in no way impairs the readability and the wealth of information available in the volume. Though this reviewer is somewhat biased toward study of myotonic dystrophy, the book really is one of the best of its genre.

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Genetic Strains and Variants of the Laboratory Mouse, 2d ed. Edited by M. F. Lyon and A. G. Searle. Oxford: Oxford University Press, 1989. Pp. 876. $175.00.

Human geneticists with a taste for the parochial often describe *Mendelian Inheritance in Man* as their bible, but mouse geneticists might apply the metaphor more aptly to *Genetic Strains and Variants of the Laboratory Mouse.* Both books are authoritative references, almost always found on the shelves or desktops of their respective practitioners. Both books also contain genetic and cytologic maps and catalogs of mutant loci—tantamount, essentially, to the recorded history of genetic variation. However, the human geneticist has other books to which to refer—for example, for details regarding chromosomal aberrations in cancer or for clinical descriptions of dysmorphic syndromes—but when it comes to mice, *Genetic Strains and Variants of the Laboratory Mouse* has it all! Where else, for example, could one find information about neuropathology in various mutations of cerebellar function, which hybridization probe to use for detecting molecular clones of human DNA in a library constructed from a mouse/human hybrid, the putative genetic mechanisms involved in Cattanach "translocation," or potential mouse models for Greig cephalopolysyndactyly.

The format of this new, second edition will be familiar to those who have a copy of the first edition, published 9 years ago. Many of the same authors contribute chapters dealing with their respective fields of expertise, but most of these sections have been considerably updated. In addition, there are new sections on wild mice and strains derived therefrom, repetitive DNA families, DNA polymorphisms, linkage and synteny homologies to man, the *t*-complex, and retroviral and cancer-related genes.

A comprehensive review of such a major reference work would be futile (and possibly presumptuous), and I will attempt instead to briefly describe its contents and to emphasize a few points which may be relevant to readers of the *Journal.* After an introductory chapter on mouse gene nomenclature, there is a large catalog of mutant genes and polymorphic loci, with a brief annotated description for each entry. A table, at the beginning of the chapter, which classifies loci by their phenotypic effects will be of enormous help to those of us searching for mouse models of human diseases, or vice versa.