chromosomes 1 and 11 in six kindreds. We are concerned about the possibility of over diagnosis of Marfan syndrome within the kindreds presented. In the six families, one would expect 37 out of the 74 offspring who are at a 1 in 2 risk of inheriting the gene to be affected. Fifty-seven were in fact diagnosed as having Marfan syndrome; this is significant ($\chi^2=5{\cdot}3$, $p<0{\cdot}05$). The disparity is even more marked when families are sampled selectively. In family 4 a lod score of 0·92 was obtained using one of the probes, D1S7. However, out of 13 subjects at a 1 in 2 risk, only two were diagnosed unaffected.

Such methodological problems, we believe, may lead to false conclusions regarding exclusion of loci which may possibly be involved in Marfan syndrome.

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BOOK REVIEW

Third International Conference on Osteogenesis Imperfecta. Ed G Cetta, F Ramirez, P Tsipouras. (Pp 187.) New York: The New York Academy of Sciences. 1988.

This volume contains papers presented at the Third International Conference on Osteogenesis Imperfecta (Pavia, Italy, 1987). Contributions on clinical, genetic, biochemical, and molecular aspects are presented, and aim to give a broad overview of current knowledge about this heterogeneous and complex group of connective tissue disorders.

Part I on nosology and genetics contains four papers discussing clinical classification and correlation of phenotype with biochemical and molecular pathology. The first paper by Sillence outlines his generally used clinical classification and contains useful radiographs. This is followed by a different and somewhat conflicting classification by Maroteaux *et al* based on antenatal or postnatal presentation, while Beighton *et al* report linkage data on families categorised according to the Sillence classification. It is clear from these papers that the biochemical and molecular heterogeneity within each defined clinical phenotype is considerable and not yet well understood.

Part II contains eight papers on biochemical defects detected in type I collagen, ranging from reports of abnormalities documented in single families to studies of substantial numbers of patients with all clinical types of osteogenesis imperfecta. No summary of collagen biochemistry is given, and a basic knowledge in this area is required in order to interpret the findings presented.

Part III contains three very good papers on the structure of the type I collagen genes, including an excellent review by Byers *et al* on the molecular basis of clinical heterogeneity. As in the biochemical papers, it is assumed that the reader will be familiar with molecular terminology.

Part IV on genetic counselling incorporates only two papers. The first by Sykes and Ogilvie concentrates on the potential application to prenatal diagnosis of linkage with either COL1A1 or COL1A2 loci in dominantly inherited OI. The paper by Thompson *et al* gives excellent practical guidelines for counselling in cases of perinatally lethal and progressively deforming osteogenesis imperfecta.

Part V on management contains four papers, three of which report experience with orthopaedic treatment from units with a special interest in osteogenesis imperfecta.

As is inevitable in a book of this format, the style and quality of the papers varies considerably, and there is a certain amount of repetition between various chapters. The fact that English is not the first language of some authors detracts from a few of the chapters. It is difficult to define the readership for this book. Scientific investigators working in this field will inevitably find certain areas already out of date, and clinicians involved with osteogenesis imperfecta will find limited new information that is not already available to them. The lack of general introductory chapters on collagen biochemistry and molecular studies detracts from the value of the book for readers who are not already conversant with these aspects. Paediatricians who are occasionally faced with the task of clarifying the diagnosis and prognosis in affected neonates would find the clinical classification useful, despite the limitations imposed by the unresolved problem of heterogeneity in this group of disorders. Despite these reservations, this book would be a useful addition to postgraduate libraries, even if few clinicians or scientists wish to purchase personal copies.

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Third International Conference on Osteogenesis Imperfecta

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