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plification and sequencing are shown in the figure. A first strand cDNA synthesis kit (Pharmacia) with poly-T primer and specific COX8 primer (F 1–22) was used for cDNA amplification between nucleotides 22 and 472 (figure A), according to the nucleotide numbers reported by Rizzuto *et al.*[7] The 5′ end of COX8 cDNA, before nucleotide 22, was amplified with the 5′-RACE kit (Life Technology/Gibco-BRL) and specific COX8 primer (R 231–250, figure B). Overlapping PCR fragments were sequenced using the dideoxynucleotide chain termination method with fluorescent dideoxynucleotides on an Applied Biosystem 373A DNA sequencer (Perkin Elmer/ABI). We found a hitherto unidentified sequence of 42 nucleotides at the 5′ end of the cDNA, and the rest of the COX8 cDNA sequence was identical to that previously described by Rizzuto *et al*[7] (figure). From nucleotides −42 to +472, there were no mutations in either the affected or healthy people in the COX8 cDNA.

The promoter region of the COX8 gene, the part of the gene that has not yet been identified, was not analysed. However, all the mutations so far identified in FHC have been described, either in the coding regions of the gene (mutations in MYH7 encoding the β myosin heavy chain[3]), or in an intronic splicing site (mutation in the splice donor sequence of intron 15 of the cardiac troponin T gene[5]). By analogy with all the data available for FHC, we therefore conclude that the COX8 gene is very unlikely to be the disease gene of the CMH4 locus responsible for FHC.

We are indebted to the family members for their invaluable participation. We are also grateful to J Weissenbach, P Millasseau, C Cruaud, and C Caloustian for their help in sequencing at the Généthon (Evry, France) and to L Fetler, O Dubourg, A Hagege, J B Bouhour, and R Isnard for their contribution to clinical diagnoses. This work was supported by INSERM (Réseau de recherche clinique No 492010) and the Association Française contre les Myopathies. GB is recipient of a grant from the Foundation Bettencourt-Schuller.

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BOOK REVIEWS

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Assessing Genetic Risks: Implications for Health and Social Policy. Editors L B Andrews, J E Fullarton, N A Holtzman, A G Motulsky. Committee on Assessing Genetic Risks, Division of Health Sciences Policy, Institute of Medicine. (Pp 338; £36.95) Washington, DC: National Academy Press. 1994. ISBN 0-309-04798-6.

This fascinating report provides a comprehensive account of the "promises and problems in genetic testing". The deliberations of the 20 committee members were informed by a series of papers and discussions at workshops, meetings, and a public forum.

The first 28 pages comprises the Executive Summary and provides the reader with a useful synopsis of the 70 plus recommendations. Many of their recommendations mirror those found in the Nuffield report on ethical issues in genetic screening (London: Nuffield Foundation, 1993). One example is the creation of a National Advisory Committee Working Group on Genetic testing to oversee professional practice and determine when new genetic tests are ready for wide scale use in medical practices. The Nuffield report proposes the setting up of a central coordinating body to review genetic screening programmes and monitor their implementation and outcome.

The report covers the following issues: Genetic testing and assessment; Laboratory issues in human genetics; Issues in genetic counselling; Public education in genetics; Personnel issues in human genetics; Financing of genetic testing and screening; Social, legal, and ethical implications of genetic testing; and Research and policy agenda.

The numerous references for each of these topics are situated at the end of each chapter. The report continuously emphasises the need to respect the autonomy of people in the way they use genetic information.

I was delighted to see two pages devoted to "Recognizing Social and Cultural Differences". The report identified the need for a variety of information and education on genetics, with balanced descriptions, in a culturally acceptable manner and at an appropriate time. I found the term "teachable moment" a very useful concept (that is, when the person is most able to comprehend the full significance of the information). Recommendations about developing innovative information materials such as interactive computer systems were noted on several occasions. As in the Nuffield report, reference is made to the training needs of primary care practitioners.

It is of interest that the Chairman of the Committee (A G Motulsky) felt the need to add a separate note to the Preface. In it he pointed out that while the majority of the committee favoured voluntary participation in neonatal screening, a minority felt that mandatory screening for phenylketonuria (PKU) and hypothyroidism would be a simpler solution.

On a more personal note, he stated that information about sickle cell trait that is incidentally detected in neonatal screening is difficult to withhold and should be given to the mother with appropriate genetic counselling. This seems to be in contrast to the more confusing recommendation of the report: "When carrier status may be incidentally determined in newborn screening (eg, in sickle cell screening), parents should be informed in advance about the benefits and limitations of genetic information, and that this information is not relevant to the health of their child. If they ask for the results of the incidentally determined carrier status for their own reproductive planning, it should be communicated to them in the context of genetic counselling, and they should be informed that misattributed paternity could be revealed."

This has been a most enjoyable book to read and I would strongly recommend it to anybody interested in the broader issues raised by genetic screening and testing.

ELIZABETH N ANIONWU

Genetics in Neurology. Bailliere's Clinical Neurology. Editor A E Harding. (Pp 452; £27.50) UK: Harcourt Brace. 1994.

How much of this rapidly expanding field can you cover in 452 pages (including references and index)? To their credit, the editor and authors of this text have included all the inherited neurological diseases which are likely to be of major interest to clinical neurologists and clinical geneticists. In addition, judging by the number of times this reviewer's copy was borrowed, molecular geneticists working with these diseases will also find the chapters enlightening.

The chapters are all well written, comprehensible, and, in addition to providing excellent reviews of their subjects, provide an insight into the issues of genetic and phenotypic heterogeneity as they relate to the specific disease groups. The chapters on hereditary ataxias (Banfi and Zoghbi) and

human prion diseases (Collinge and Palmer) in particular show how we are only just beginning to understand the relationships between genotype and phenotype.

From the point of clinical applicability, the discussion, in the chapter on the muscular dystrophies (Bushby), on the use of dystrophin DNA and protein analyses in suspected Xp21 dystrophy patients, carriers, and at risk pregnancies, is likely to be of particular interest.

In addition to the expected topics of Alzheimer's disease, prion diseases, movement disorders, ataxias, neuropathies, neuronopathies (spinal muscular atrophies), mitochondrial diseases, muscular dystrophies, and myotonias three chapters cover disorders where the genetic component is less well understood, the epilepsies, multiple sclerosis, and neurological tumours. The last of these does include brief discussions on the genetic neurocutaneous syndromes but it deals largely with the molecular genetics of sporadic neurological tumours. These are too often omitted from texts discussing neurological genetics, and this chapter provides a valuable detailed discussion on the roles of chromosome loss and gene mutations (p 53) in specific CNS tumours.

All of the 12 chapters are as current as the publication date of August 1994 allowed. Indeed the only major additions/updates a clinical neurologist is likely to need are Machado-Joseph disease, which was recently appended to the list of trinucleotide repeat expansions affecting the CNS, the gene for Emery-Dreifuss muscular dystrophy, and the gene(s?) for the autosomal recessive spinal muscular atrophies of childhood.

There appear to be very few typographical errors, the most notable being the substitution of heterozygotes for homozygotes at one point in the description of the modifying effect of codon 129 status on age at onset for inherited prion diseases (p 248).

Overall this volume has much to commend it, including the price. It should certainly be read once by clinical geneticists (and neurologists) especially if they feel the need to "catch up" on the recent past advances in the molecular genetics of the major groups of inherited neurological diseases.

JOHN MACMILLAN

Textbook of Fetal Physiology. Editors G D Thorburn, R Harding. (Pp 468; £65.00.) Oxford: Oxford Medical Publications. 1994. ISBN 0198577486.

The preface to this excellent volume states that it aims to act as a companion to the classical texts on adult physiology for medical training at undergraduate and postgraduate levels. In fact it fills an important niche in the market owing to the paucity of readable books on human fetal development and physiology. Indeed there is also a rapidly increasing demand for this type of general textbook from human molecular geneticists. As the combined efforts of human genome project related research generate an ever expanding list of cloned genes associated with specific human diseases, the task remains of elucidating how any gene product actually causes the associated disease. Inevitably questions of gene expression during development arise. The textbook of fetal physiology will provide a valuable reference book for molecular and cell biologists asking these questions.

The book has chapters from a large number of contributors on topics ranging from placental function, through development of individual organ systems and physiological mechanisms, ending with chapters on maternal aspects of pregnancy. For each organ system a first chapter addresses the embryological, structural, and cellular aspects of development. One or more subsequent chapters then address an important physiological function of that organ system. Hence, the chapter on development of the respiratory system is followed by a chapter on fetal lung maturation, and that on renal function is accompanied by one on fetal fluid balance. There is a particularly extensive section on different aspects of development of the nervous system. The book is well illustrated and readable and will undoubtedly be a useful addition to many medical and basic science bookshelves.

ANN HARRIS

NOTICE

Gene Therapy

The 3rd International Karger Symposium on Gene Therapy will be held on 22–24 October 1995 in Basel, Switzerland. For further information contact: S Karger AG, Congress Secretariat, PO Box, CH-4009 Basel, Switzerland. Tel: +41/61/306 11 11, fax: +41/61/306 14 34.

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The readership of *Journal of Medical Genetics* is world wide and covers a broad range of workers, including clinical geneticists, scientists in the different fields of medical genetics, clinicians in other specialities, and basic research workers in a variety of disciplines. It publishes original research on all areas of medical genetics, along with reviews, annotations, and editorials on important and topical subjects. It also acts as a forum for discussion, debate, and information exchange through its Letters to the Editor columns, conference reports, and notices. The editor is always grateful for suggestions or criticisms from readers and authors.

ORIGINAL PAPERS

These may be on any aspect of medical and human genetics and may involve clinical or laboratory based and theoretical genetic studies. If requested, authors shall produce the data upon which the manuscript is based for examination by the Editor. Guidance on length can be obtained from studying the Journal. Case and family reports may be submitted as *Brief papers*. *Short reports* should in general not exceed 500 words, with one or two illustrations, and the text should be continuous with no headings. An abstract should be provided for all papers. Contributions may also be submitted as *Hypotheses* or *Technical notes*. Accelerated publication of papers of particular importance will be considered.

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Short or longer reviews on all aspects of medical genetics are welcome, but should be discussed first with the Reviews Editor. Contributions on historical topics, or which could form part of specific series, are particularly acceptable.

ANNOTATIONS AND EDITORIALS

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LETTERS

These are welcome on any relevant topic and will be published rapidly. Those relating to or responding to previously published items in the Journal will be shown to those authors, where appropriate. Although a paper submitted as an original report may sometimes be published in shortened form as a letter, it is preferable for initial submissions to be as a short report, unless directly related to a previous journal article.

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BOOK REVIEWS

The Journal aims to review as wide a range of relevant books as possible. Authors or others wishing to check if a book has been received may check with the Journal office. Computer programs and databases, official reports, and other material relevant to the field may all be appropriate for review. Enquiries about such items are welcome.

OBITUARIES

The Journal would like to be informed rapidly of the death of any senior or important person in the field of medical or human genetics, regardless of geographical location. In general, a brief notice would be published rapidly, with a longer obituary as appropriate. Since such deaths often occur many years after retirement, it will be appreciated if readers will contact the Reviews Editor so that appropriate arrangements can be made.

NOTICES

Notice of forthcoming meetings in different countries should be sent as far ahead as possible. Extensive descriptions should be placed as advertisements.

'CALLS FOR PATIENTS'

The Journal receives an increasing number of requests to publish notices of proposed studies involving patients or families with rare genetic disorders. In general such notices are appropriate only for major international collaborations; the proposer should ensure that such a notice does not conflict with existing studies or proposals.

ILLUSTRATIONS

High quality black and white photographs are preferred for most illustrations, particularly of patients. Colour illustrations can be accepted; however, authors are asked to pay part of the cost, so their desirability should be discussed in advance of submission. All identifiable photographs of patients must be accompanied by written permission for use.

NOTES ON NOMENCLATURE

Authors should refer to the following publications.

(1) Chromosomes: *ISCN 1985. An international system for human cytogenetic nomenclature*. Basel: Karger, 1985.

(2) Genes: Shows TB, *et al*. In: Human Gene Mapping 5 and 7. *Cytogenet Cell Genet* 1979;**25**:96–116, 1984;**37**:340–3.

(3) Loci: Conventional nomenclature should be used, with lower case lettering as appropriate (for example, Race RR, Sanger R. *Blood groups in man*. 6th ed. Oxford, London: Blackwell, 1975; and Giblett ER. *Genetic markers in human blood*. Oxford, London: Blackwell, 1969).

(4) Blood coagulation: International Committee of Haemostasis and Thrombosis (Graham JB, *et al*). A genetic nomenclature for human blood coagulation. *Thromb Haemostas* 1973;**30**:2–11.

(5) Enzymes: *Enzyme nomenclature: recommendations of the nomenclature committee of the International Union of Biochemistry*. New York: Academic Press, 1984.

Specific instructions to authors

Papers, which should be in triplicate and in the Vancouver style (*BMJ* 1988;**296**:401–5), should be sent to the Editor, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR and not to individual editors, with the exception of papers from the USA, which can be submitted to the North American Editor, Dr P M Conneally, Department of Medical Genetics, James Whitcomb Riley Hospital for Children RR129, Indiana University Medical Center, Indianapolis, Indiana 46223, USA. Submission of a paper will be held to imply that it contains original work which has not been previously published. It is the responsibility of the submitting author to ensure that all co-authors are agreeable for their names to appear on the manuscript. A FAX number should be provided. Permission to republish must be obtained from the Editor.

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References should conform precisely to the style current in this journal. Authors are responsible for the accuracy and completeness of their references as these will not be checked by the Editorial office.

Up to four keywords should be provided for indexing purposes.

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