

## LETTER TO THE EDITOR

### Grebe syndrome: a second case with extremely severe manifestations

We read with great interest the report by Kulkarni *et al*<sup>1</sup> on Grebe syndrome in a severely affected child. We have recently seen a similar case, a 28 week preterm female infant with a birth weight of 600 g and a length of 25 cm, who died soon after birth (fig 1). She was the first child of a consanguineous couple (first cousins once removed) of Italian ancestry. The family history was unremarkable and the pregnancy had been uneventful. Clinical examination showed marked micrognathia, a normal neck and trunk, and shortening of all four limbs, the lower being more severely affected than the upper. The shortening was progressive, from proximal to distal, the fingers were bud-like (fig 2), the lower limbs very short, the hips abnormally jointed, and there was no clear limit between the thighs and legs. Distally, close to the ankles, there were osseous spurs protruding bilaterally. The feet were internally rotated and the toes, as well as the fingers, were knob-like (fig 3).



Figure 1 Full body view.

The radiographs showed normal humeri, absent ulnae, and very poorly ossified metacarpals and phalanges. Both femora were absent and, distally, there was only an unidentifiable rudimentary bone, the one observed clinically as a bony protrusion. Again, the metatarsals and phalanges were very poorly ossified or not at all. The skull, spine, ribs, and pelvis were normal (fig 4). No internal malformations could be detected at necropsy and cartilage histology was normal. Neither of the parents showed any clinical abnormality.

Because of the lack of trunk involvement and the radiographically normal bone struc-



Figure 2 Upper limbs. Note bulbous digits.

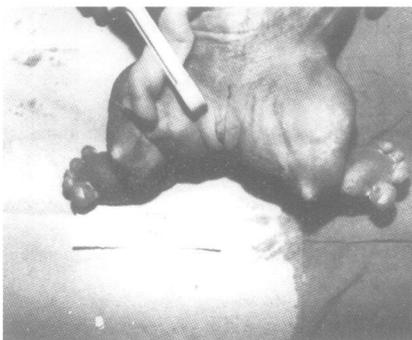


Figure 3 Lower limbs. Note bony spurs and bulbous toes.



Figure 4 Whole body radiograph. Note absence of femora and ulnae and only one bone in the legs bilaterally.

ture, lethal skeletal dysplasias were excluded as differential diagnoses, as were femoral hypoplasia-unusual facies<sup>2</sup> and femur-fibula-ulna<sup>3</sup> syndromes. The whole clinical and radiographical appearance suggested Grebe syndrome.

Grebe syndrome is a rare genetic entity, which to date to our knowledge has been described in about 70 cases, belonging to 12 families or inbred groups. There seems to be no doubt about its autosomal recessive mode of inheritance, based on recurrence in sibs or parental consanguinity or both. Although the overall clinical picture in the different sib-

ships described was similar, the slight variations among them, as well as minor expression in heterozygous carriers mentioned by some of the authors,<sup>4,5</sup> could indicate that different genetic entities or perhaps different mutations of the same gene are involved.

Our patient seems to be the second case with an extremely severe form of this condition, which could, as Kulkarni *et al*<sup>1</sup> suggested, reflect full expression of the gene, or, as already mentioned above, indicate that a different genetic entity or a different mutation of the same gene could be involved. Further studies on the recently described DCMP1 mutation<sup>6</sup> apparently involved in Grebe syndrome should clarify this question.

M RITTLER

Medical Genetics Unit, Hospital Materno Infantil  
Ramón Sardá, Buenos Aires, Argentina

S HIGA

Department of Pathology, Hospital Materno Infantil  
Ramón Sardá, Buenos Aires, Argentina

- 1 Kulkarni ML, Kumar B, Nasser A. Grebe syndrome: a very severely affected case. *J Med Genet* 1995;32:326-7.
- 2 Burn J, Winter RM, Baraitser M, Hall CM, Fixsen J. The femoral hypoplasia-unusual facies syndrome. *J Med Genet* 1984;21:331-40.
- 3 Tentamy S, McKusick V. Femur-fibula-ulna syndrome. In: Bergsma D, ed. *The genetics of hand malformations*. New York: Alan R Liss, 1978:155-6.
- 4 Quelce-Salgado A. A rare genetic syndrome. *Lancet* 1968;i:1430.
- 5 Curtis D. Heterozygote expression in Grebe chondrodysplasia. *Clin Genet* 1986;29:455-6.
- 6 Thomas JT, Kilpatrick MW, Lin K, *et al*. Disruption of human limb morphogenesis by a dominant negative mutation in CDMP1. *Nat Genet* 1997;17:58-64.

## BOOK REVIEWS

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**Chromosomes Today.** Volume 12. Editors N Henriques-Gil, J S Parker, M J Puertas. (Pp 379; £60.00.) UK: Chapman & Hall. 1996. ISBN 0-412-75240-9.

This book contains all the invited lectures given at the 12th International Chromosome Conference held in September 1995 in Madrid, Spain. Not surprisingly, fluorescence in situ hybridisation has added colour to the classical beauty of chromosomes. The reader is attracted by a colourful cover and many colour plates. Indeed much progress has been made by using the new molecular cytogenetic techniques to study the complexities of chromosome structure, function, and evolution. However, I couldn't agree more with Peter Moens saying that "chromosome images have become exquisite but the line between fact and fancy is sometimes treacherously narrow", but he also states clearly that "nostalgia

has no place in science". I found the prologue of this book, a short history of cytogenetics by Lacadena, and the epilogue, which gives Moens's personal views on the Chromosome Conference(s), most readable. Here you will certainly find some citations and bonmots for lectures and chromosome courses.

The various sections of this book deal with chromosome organisation, chromosome stability, control mechanisms, meiosis, and evolutionary dynamics. In general, the articles are of high scientific quality. However, their length and format are quite different. Some chapters, for example on "Y-linked genes and spermatogenesis in primates" by Schempp and on "gametic imprinting at the mouse and human IGF2R/MPR300 locus" by Smrzka and Barlow, are quite brief and read like short reports. Other chapters give more background information. In this regard, I found the chapters on "the relationship between gene density and chromosome banding patterns in mammalian nuclei" by Craig and Bickmore and on "the replication of ribosomal RNA genes in eukaryotes" by López-Estraño *et al* to be excellent. Peter Cook describes a minimalist chromosome model which does not involve solenoids and helical coiling. Instead transcription factories are thought to be the precursors of mitotic chromosome bands. Since by the time this book has come out, most of the original data presented have already been published elsewhere, I prefer these review-like and sometimes unconventional papers.

A book on chromosomes should be interesting for a wide variety of chromosomologists and this conference report certainly is. It impressively shows that molecular biology has been incorporated into cytogenetics, and yet that classical cytogenetic techniques still play an important role. It is not unexpected that the various chapters describe the chromosomes of widely different species ranging from plants to *Drosophila* and other insects, as well as of mammals, primates, and humans. The very concise papers on "retrotransposons at *Drosophila* telomeres and terminal chromosome deficiencies" by Biessmann *et al* and on "proteins controlling sister-chromatid cohesion" by Orr-Weaver *et al* teach us why the fly is still a highly valuable model organism. What seems to me to be somewhat neglected is yeast, an organism for which cytogenetic methodologies are beginning to be established. A few paragraphs of Scherthan's paper on "chromosome behaviour in earliest meiotic prophase" give at least some insight into the power of yeast cytogenetics.

Overall the topics range from the esoteric, for example, on "chromosome differentiation using nucleases" by Gosálvez *et al* and on "evolution of a near-neutral B chromosome" by Camacho *et al*, to the highly practical. (By

necessity, this categorisation is highly biased.) For the human geneticist, the latter category includes good chapters on "subchromosomal painting libraries from somatic cell hybrids" by Rocchi *et al*, on an "interspecific micronucleus model for the study of induced chromosome aberrations in human male germ cells" by Egozcue *et al*, and on the "characterization of amplified DNA sequences in human cancers" by Muleris *et al*. Perhaps even more impressive are the two articles on "chromosome painting in wheat" by Vega *et al* and on "new insights into chromosome evolution in plants from molecular cytogenetics" by Leitch *et al*, which elucidate the immense agricultural value of the new cytogenetics.

In summary, this book is a very valuable source of information for chromosome researchers or, more generally speaking, for people who love chromosomes. It is part of a larger and well known series of conference reports which should be found on the shelves of every genetic institute. Since not all chromosomologists can afford to attend the International Chromosome Conference, a premier event in our field, as many as possible should get a chance to browse through this book. It is both informative and amusing.

THOMAS HAAF

**Genes VI.** Benjamin Lewin. (£29.25.) Oxford: Oxford University Press. 1997. ISBN 0-19-857778-8.

For a book to reach its sixth edition in 15 years in the competitive field of molecular genetics textbooks, it has to be good. Though lecturers and tutors may put specific textbooks on reading lists, in my experience the students have their own "word of mouth" ranking for these general texts. It is easy to see why Lewin still attracts the buyers. The text is clearly written, comprehensive, and, most importantly, exciting. Some aspects of molecular genetics are inherently interesting, for example, what switches a gene on and off in differentiated cells of the body. Other topics are more difficult to present in a way that keeps the imagination alert, but Lewin succeeds admirably. The text of *Genes VI* has been rearranged and updated as the subject has evolved. This edition is divided into seven main parts, covering DNA; genes to proteins; prokaryotic gene expression; perpetuation of DNA; eukaryotic genomes; eukaryotic gene expression; and a final section on cell growth, cancer, and development. Each section is packed full of information, making this an extremely useful reference book as well as a teaching textbook. In fact, my only criticisms of *Genes VI* are presentational: the rather stark colour scheme of figures gets rather tir-

ing after a while and the weight of the book, which approximates the Yellow Pages for a medium sized city in England, is not really suitable for the rather flimsy softback version. The production team needs to consider this for future editions, or maybe these will only be available on CD-ROM.

ANN HARRIS

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## NOTICES

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### Medical Editors Trial Amnesty

A large number of internal journals have joined in a Medical Editors Trial Amnesty to encourage those who have controlled trial data, which have not been fully published, to bring such data into the public arena. This is an important effort to minimise publication bias. It is relatively unlikely that this journal will be the most suitable for such publications but should there be any unreported trials that are suitable for the *Journal of Medical Genetics*, the editors would be pleased to consider them. More comprehensive details will be publicised in editorials in other clinical journals.

### Genetic Aspects of Autoimmune Diseases

A conference for scientists and representatives of patients' organisations on "Genetic Aspects of Autoimmune Diseases" will be held on 7-10 May 1998 at Noordwijkerhout, The Netherlands. For further information contact Mrs E M Kalsbeek, WOCZ, PO Box 8459, 3503 RL Utrecht, The Netherlands. Tel: +31 30 2966400, fax: +31 30 2970020, e-mail: conference@wocz.spin.nl

### Enzymes, Receptors, and Drugs in Obesity and Atherosclerosis

An international symposium on "Enzymes, Receptors, and Drugs in Obesity and Atherosclerosis" will be held on 7-9 May 1998 at the University of Toronto, Toronto, Ontario, Canada. For further information contact Dr H V Markle, Department of Laboratories, Centenary Health Centre, 2867 Ellesmere Road, Scarborough, Ontario, Canada M1E 4B9. Tel (416) 281-7251, fax: (416) 265-8781, e-mail: HVM@myna.com Web site: <http://www.clinitox.com/erdoa/>

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## Correction

In the August 1997 issue of the Journal, in the review of the book *Genetic Disorders Among Arab Populations* on page 704, the price of the book should have been \$98.50 and not \$498.50.

# Referees, 1 September 1996-30 September 1997

Those listed below have contributed their time and expertise to review articles for the journal. Their help, in maintaining high standards of publishing, is enormously appreciated.

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