Nilbert *et al*[3] reported a uterine leiomyosarcoma with a t(8;13). In addition, several uterine leiomyomas with structural or numerical abnormalities of chromosome 8 were reported by Mark *et al*[4] and Teyssier and Ferre.[5] Especially interesting in relation to the case under discussion is the latter authors' report of trisomy 8 in another gastrointestinal smooth muscle tumour, an oesophageal leiomyoma.

Thus, the abnormalities in chromosome 8 in smooth muscle tumours described so far involve both numerical and structural abnormalities and concern both benign and malignant tumours. We think that these data give a different perspective to the discussion of the case reported by Lessick *et al*.[1]

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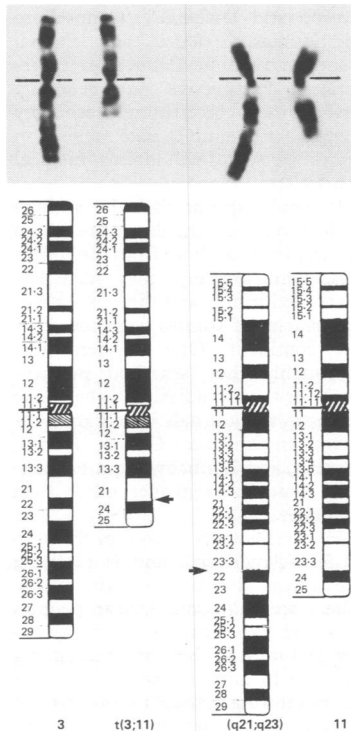
Further evidence for the location of the BPES gene at 3q2

We read the paper of Smith *et al*[1] in this journal with interest. They suggested that blepharophimosis plus ovarian failure is a likely candidate for a contiguous gene syndrome, and recommended cytogenetic investigation of all cases of blepharophimosis, ptosis, epicanthus inversus syndrome (BPES). We would like to report a family with autosomal dominant BPES syndrome and a chromosomal abnormality. A father and his 6 month old son were referred for genetic counselling. Both showed the typical signs of BPES: blepharophimosis, ptosis, telecanthus, and epicanthus inversus. The father had no other dysmorphic features and was of normal intelligence. The son had a small nose with anteverted nostrils and cup shaped ears. His height, length, and head circumference were in the normal range and his mental development was normal. The father had two sons from a previous marriage who had the same eye anomalies. Unfortunately, they were not available for further investigations. Chromosomal examination on cultured lymphocytes of the father and son showed an apparently balanced translocation between the long arms of chromosomes 3 and 11, with respective breakpoints at 3q21 and 11q23. The karyotype was 46,XY,t(3;11)(q21;q23) (figure).

Recently, Fukishima *et al*[2] reported a newborn infant with BPES and a de novo balanced 3q23;4p15 reciprocal translocation. These findings strongly indicate that the gene for BPES is located in the 3q2 region. Furthermore, blepharophimosis, ptosis, and microphthalmia are consistent features in patients with an interstitial deletion of band 3q2,[3] reinforcing the location of the BPES gene at 3q2.



Karyotype of the proband.

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BOOK REVIEWS

All titles reviewed here are available from the BMJ Bookshop, PO Box 295, London WC1H 9TE. Prices include postage in the UK and for members of the British Forces Overseas, but overseas customers should add £2 per item for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank, or by credit card (MASTERCARD, VISA, or AMERICAN EXPRESS) stating card number, expiry date, and your full name.

Pathology of the Human Embryo and Previable Fetus. An Atlas. D K Kalousek, N Fitch, B A Paradice. (Pp 230; DM 248.) Berlin: Springer-Verlag, 1990.

This is a beautifully produced book by experts for experts. There are not

enough experts in the field of embryo/feto pathology and this atlas may well be the starting point to awaken the interest of pathologists, medical geneticists, and ultrasonographers in this important field.

The book is divided into three parts. The first part concerns embryonic and fetal development, the second part the examination of aborted embryos and fetuses, and the third part discussion of pathological disorders in the embryo, fetus, and in placental development. There are useful appendices giving suggestions for protocols, consultation forms, and some detailed measurements.

The first part of the first section is perhaps one of the weakest points. Standard diagrams are used to illustrate the very early stages of development and here I think it would have been good to have some of the marvellous scanning electron microscopic photographs which are now available. However, the book does not set out to be an embryological text and this should in no way be seen as detracting from its general excellence. Later photographs and diagrams of older embryos and fetuses are excellent, and I thought reproduced well in black and white until I saw the small selection of colour plates at the end of the book which show detail even better. I imagine that producing the book all in colour would have made it prohibitively expensive and the black and white illustrations serve their function.

The section which deals with pathology in the embryo and fetus is superb and reflects the authors' extensive experience in this field. Practical techniques for examination of the embryo and fetus at all stages are given and all through the book there is emphasis on correct terminology and conventions about currently accepted categories. This is most important since it avoids a person newly introduced to the field reinventing the wheel.

In summary, this is a splendid book which all pathologists who are involved in fetal examination should have in their own departments. All genetics departments involved in fetal examination and fetal dysmorphology should own a copy and I think it should be in all university medical libraries.

D DONNAI

Genetic Variation and Disorders in Peoples of African Origin. James E Bowman, Robert F Murray. (Pp 472.) Baltimore: The Johns Hopkins University Press. 1990.

This book is a veritable compendium of genetic information, largely on American blacks but also on various African populations, systematically arranged for easy reference. The emphasis is on normal variation but the authors have tried valiantly to relate this variation to disease. In the case of haemoglobin and G6PD variation this is relatively easy and has been well done. When it comes to other serological polymorphisms they have, understandably, been less successful. The chapters on 'Anthropometry and Skeletal Variation' and on 'Dermatoglyphics', which together constitute 18% of the length of the book, have very little relevance to health and will not be of much interest to medical geneticists. Those chapters on the malaria protective traits, including one devoted to 'The Malaria Hypothesis', and the chapters on 'Lactose Intolerance and Malabsorption', 'Twins and Other Multiple Births', 'Congenital Malformations', 'Hypertension and Diabetes', 'Counselling and Human Genetics', and 'Ethical Issues and Public Policy: An International Perspective' are generally well written, constituting reasonably comprehensive, up to date reviews on the subjects, and will interest medical geneticists.

It would appear that the authors wished to review the data from Africa as completely as those from the Americas. Unfortunately, they have not succeeded in this and there is a striking paucity of studies from the African continent. One wonders how thoroughly they searched publications for these studies! Did they have access to the journals which are published in Nigeria, Ghana, Kenya, Zambia, and Zimbabwe, not to mention those in languages other than English?

Hypertension is a major problem in African–Americans, and is plaguing the urban dwellers of Africa too. There are still some African peoples in whom hypertension is not a problem and most of these are undergoing rapid sociocultural change. The careful monitoring of these for the onset of hypertension, together with the documentation of their change in life style, might provide important clues as to the aetiological factors involved in the development of hypertension.

The inequalities in health between black and white Americans are clearly spelled out; with respect to almost every indicator of health, blacks fare worse than whites. Life expectancy at birth is five to six years shorter; the infant mortality rate is almost twice as high; the maternal mortality rate is over three times higher. Heredity cannot be said to be the cause of these differences; the problem is clearly socioeconomic with poverty the root cause. It is salutary to read that, whereas 9·7% of white families in the USA live below the poverty level, a staggering 32·4% of blacks do so. The plight of the Midwestern blacks is the greatest. Turning to the health of Africans, the overriding importance of environmental agents in the causation of disease is clearly brought out.

Where infant mortality rates are almost everywhere nearly 100 (and in some countries they are over 200!), it is understandable that inherited disorders have not attracted the serious attention of governments and health planners. The role of heredity in susceptibility to infectious disease is not discussed except in one case, that is, the allele *GclF* in the group specific component or Vit D binding protein system and the claim that it predisposes to AIDS (p 341). The 1987 study claiming to have found this predisposition was, we know, quickly challenged and was eventually retracted by the authors (Eales *et al*, *Lancet* 1988; i:936). The spurious association was the result of an unfortunate laboratory error and it is a pity that the correction, published in April 1988, is not referred to in this book which carries a 1990 date!

The chapters on 'Genetic Counselling and Its Adaptation to Varying Needs' and 'Human Genetics, Ethical Issues and Public Policy: An International Perspective' are excellent and provide fine overviews of the problems as they affect First World countries and, in particular, the socioeconomically privileged citizens of such countries. There is little attempt, however, to grapple with the problem of the role of genetic services in developing countries. In a brief discussion of the economics of genetic services in the United States, the authors quote, apparently with approval, the recommendation that "administrators