Editorial

Journal of Medical Genetics, 1984, **21,** 404–406

Lateral reading 9

GENETICS, DYNAMICS, AND POLITICS (Lawton JH, May RM. *Nature* 1984;**309**:744–5)

John H Lawton and Robert M May give a report on the Dahlem conference on 'The exploitation of marine communities' held in Berlin on 1 to 6 April 1984. Two interesting genetic possibilities may result from commercial fishing.

(1) In fishing a previously unexploited stock there is often faster growth and earlier breeding of the surviving population, generally attributed to the fact that as stocks are fished down food supplies per individual increase, leading to faster growth and earlier maturity. However, there is also a genetic possibility if growth rates and age of maturity are heritable. In this case fishing would represent a massive novel selection on the population. Individuals that grow faster and/or mature earlier may be at an advantage in the fish population because they are more likely to reproduce before being caught.
(2) Sometimes fishing leads to a reduction in growth rates and to an increase in the mean age of fish. This happened in the case of the Lesser Slave Lake Whitefish and may be the result of heavy selection of gill nets on large rapidly growing genetic variance in a species where reproduction is strictly determined by age and not by size.

ORNITHINE DECARBOXYLASE AS A BIOLOGIC MARKER IN FAMILIAL COLONIC POLYPOSIS (Luk GD, Baylin B. *N Engl J Med* 1984;**311**:80–3)

The authors of this paper had previously shown that the onset of proliferation of rat intestinal mucosa is dependent on increased activity of ornithine decarboxylase. They therefore measured ornithine decarboxylase activity in biopsy specimens of colonic mucosa and polyps from patients with polyposis and at risk family members.

In colonic mucosa from 16 normal controls, ornithine decarboxylase activity was less than 2·5 nmol/mg per hour. In contrast, it was higher than 2·5 nmol/mg per hour in the normal-appearing areas of colonic mucosa from 11 to 13 patients with familial polyposis and in all polyps biopsied from these same subjects ($p<0·05$ for specimens from both sites, as compared with controls). Mucosa from dysplastic polyps showed higher mean ornithine decarboxylase activity than mucosa from polyps that were not dysplastic ($p<0·05$). In colonic mucosa from clinically unaffected, first degree relatives of patients with familial polyposis, there was a bimodal distribution of ornithine decarboxylase activity, with one peak at the mean for normal controls and the other near the mean for normal appearing mucosa from affected patients.

The paper, with a leader, suggests that ornithine decarboxylase activity in colonic mucosa may reflect the abnormal proliferative state in familial polyposis and identify clinically normal family members who carry the genotype.

DIABETES MELLITUS (DM) IN THE AETIOLOGY OF DUPUYTREN'S DISEASE (DD) (Noble J, Heathcote JG, Cohen H. *J Bone Joint Surg [Br]* 1984;**66**:322–5)

Dupuytren's contracture again. Of 122 randomly chosen adult diabetics DD was found in 27 of 65 men (41%) and 24 of 57 women (42%). As might have been expected the incidence of DD was related to the length of time the patients had had DM, this probably being because increasing age is the major factor in making DD manifest. The proportion of patients with DD was the same whether the diabetes was or was not insulin dependent and poor control of the DM was not a factor.

The question posed by the authors is whether the biochemical disturbance of diabetes caused DD or whether the pattern of inheritance predisposes to both DD and DM.

To me it is quite clear that DD is inherited as an autosomal dominant and, incidentally, when ascertaining it is most important to examine the feet as well as the hands. Non-insulin dependent diabetes is often also inherited as an autosomal dominant whereas insulin dependent diabetes is related to autoimmune factors and virus infections, but there is some genetic influence as judged by an association with certain HLA types.

The genes in DD and non-insulin dependent diabetes might be linked and it would be interesting to carry out family studies to see if there were crossing over. If linkage were demonstrated there would be no need to invoke a 'cause' for the 'association', it would be chance and the genes would simply be on the same chromosome.

Family studies in the insulin dependent cases with

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DD might show that there was a similar pattern of HLA antigens with DD as with DM.

The most important factor of all is to remember that 25% of male OAPs in this country have DD (the more you look for it the more you find it) and with such a common condition association with other diseases is likely to be found frequently, particularly in those who are 'getting on'.

RICHES MAKE FOR LONGEVITY (Anderson A. *Nature* 1984;**310**:92)

The life expectancy of both Japanese men and women is now the highest in the world, 78·8 years for women and 74·2 years for men. The change has been some two to four times faster than in western nations—it did not reach 50 years until 1947 and 70 until 1971—and the post war decline in population growth was so sudden that Japan is now well on the way to becoming the first geriatric society. A white paper also charts for the first time in Japan the striking relationship between life expectancy and income levels, so that riches, not genes, make for longevity.

The changes in Japanese life expectancy have largely been attained by reduction of stillbirths and all deaths during birth and the early years of life, so that the gains for those who survive these dangerous years compared, say, with someone living 100 years ago are not so great as might first appear. Nevertheless, putting the life expectancy figures together with the average age of marriage shows that the average couple might look forward to nearly 48 years of married life.

WHEN SEX IS BAD (Leader. *New Scientist,* 26 January 1984, 24)

"Sexual reproduction produces genetic variability in the offspring on which selection can operate. This means that a sexually reproducing species is more likely to adapt and survive in a changing environment than a species reproducing asexually. Now, it has been shown that the converse is also true. Asexual reproduction improves the survival chances of a species living in a stable environment.

In mutualistic symbioses one species, the inhabitant, lives largely within a second species, the exhabitant, for their mutual benefit. The two partners are exposed to quite different environments. The inhabitant, protected to a large extent from predators and pathogens, lives in a very sheltered environment. But the exhabitant is open to the same environmental stresses as a non-symbiotic species. In such symbioses the partners will have been subject to different selection pressures over many generations and will have experienced different rates of genetic change. If sexual reproduction has evolved in response to the need for variation for survival in a hostile environment then in theory the inhabitant species should reproduce asexually and the exhabitant species sexually.

R Law and D H Lewis of the Department of Botany at Sheffield University have tested this theory by examining several mutualistic symbioses and their findings in general support it (*Biological Journal of the Linnean Society,* vol 20, p 249). They found that sexual reproduction is absent or occurs rarely in most inhabitant species but occurs much more often in their free-living relatives, while exhabitant species usually reproduce sexually. The evolution of sexual reproduction does seem to have been encouraged by living in a hostile environment."

OSTEOGENESIS IMPERFECTA AFTER THE MENOPAUSE (Paterson CR, McAllion S, Stellman JL. *N Engl J Med* 1984;**310**:1694–6)

Paterson *et al* studied the fracture rate as a function of age in 45 women and 20 men with osteogenesis imperfecta. In each variant of the disorder, the fracture rate in women peaked in childhood, declined in adolescence, and rose again after the menopause. In contrast, the fracture rate in men remained low after adolescence. After the menopause women were vulnerable to crush fractures of the spine as well as fractures of the long bones.

The authors conclude that the increased fracture rate after the menopause in women with osteogenesis imperfecta reflects the superimposition of the effects of age related bone loss on those of the defective collagen structure of osteogenesis imperfecta, and that hormone replacement therapy may be specifically indicated in this group of patients from the time of the menopause. They also suggest that osteogenesis imperfecta should be included in the differential diagnosis of women presenting with crush fractures of the spine.

ABSTRACTS—FALSE SCIENCE (Editorial. *Arch Dis Child* 1984;**59**:497)

I thought this editorial from *Archives of Disease in Childhood* might be of interest.

"Authors who present papers at scientific meetings enjoy having the abstract published and seeing their name in print. It adds status to the communication and some authors include abstracts as publications on their curriculum vitae. Several long established societies have been able to secure a regular place for their abstracts in reputable scientific journals but the

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multiplication of new societies has meant that most journals are unable, or unwilling, to publish society abstracts; the result has been for abstracts to be published as separate books, or in the case of the 1983 Paediatric World Congress, as a 1000 page, three volume book, costing $50.

We do not publish abstracts (apart from a few, selected for historical interest, from the annual meeting of the British Paediatric Association) partly because we have had personal experience of preparing abstracts for meetings and recognise their limitations. All too often the abstract is written several months before the meeting, when the subject of the work is no more than an idea in the author's mind or, at best, the final results have not been analysed. These abstracts tend to be a vague and inaccurate description of the results the author is hoping for. At an international paediatric organ specialty meeting last year, study of 7 consecutive spoken presentations showed that the results presented differed from those published (as abstracts printed in a reputable European paediatric journal), in all but one of the 7. In the other 6 there were major discrepancies which made the published abstracts false. For the massive international meetings the position is even more chaotic. At the 17th International Congress of Paediatrics there was one free paper session in which only two of the 7 presentations listed were given, because the speakers did not turn up. Yet all 7 presentations appear in printed form in the official proceedings of that congress. Whether the work which was not presented was ever anything more than an idea in the authors' mind we shall never know.

A few scientific societies control and check the authenticity and value of published abstracts, but in clinical medicine such quality control is rare. Those clinical societies that attempt to exercise quality control by asking their members to vote on the suitability of the presentation for publication as an abstract tend to create merely embarrassment.

It can be useful to look through abstracts to see what workers in other parts of the world are doing. But unfortunately there is an increasing tendency for the information contained within published abstracts to be quoted as reliable information and for the reference for the abstract to be given at the end of the scientific articles. It has become increasingly apparent to us that many abstracts report work that has never been presented, which may not have been done, and which if it was done produced a different result. Because of that the editorial board has decided that it will no longer allow abstracts to be cited as reference for information contained within articles published in our journal. We are not prepared to deal in dud currency."

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C A Clarke

I have included in previous 'Lateral readings' several views about publishing abstracts. I invited comments and there follow letters from Peter Harper and Sarah Bundey.

The Editor has asked for comments on publication of abstracts, particularly those of the Clinical Genetics Society, whose presented paper abstracts are currently published in this Journal. There is no doubt that many scientists now attach a lot of importance to abstracts and regard them as publications. I do not think this is a very healthy situation and it is difficult to know how to improve things. My suggestions are:

(1) To keep abstracts really brief to avoid them turning into what is essentially a preliminary paper.

(2) Where abstracts are to be published they should have been vetted by the scientific committee of the society, not included automatically.

(3) Abstracts of reviews are generally not worth publishing.

(4) The editor of the journal should reserve the right to reject abstracts in the same way as any other publication.

This is, I think, more or less what happens currently with the Clinical Genetics Society abstracts. The one other specific point which I feel is relevant is that posters should not be regarded as an inferior category just because they are posters. They are often better than some of the contributed papers.

Peter S Harper

I write in reply to the editor's request for views on the article of Bray (*J Med Genet* 1984;**21**:163) about his concern (expressed in *Nature* 1984;**307**:206) over the increasing numbers of unrefereed abstracts which are published. I share Bray's concern. I think that often abstracts are prepared before the data have been fully analysed and may, indeed often do, contain unwitting errors. An example is the abstract in the *American Journal of Medical Genetics* 1982;**34**:90A relating to a paper in the June issue of *Journal of Medical Genetics* (1984;**21**:186–8) where the abstract gives risk estimates for genetic counselling in myotonic dystrophy that are different from those given in the full paper. I do not consider that writers of full papers should use abstracts in their list of references unless it is absolutely clear that the abstract contains sufficient data for the reader to be certain that the conclusions are well founded.

Sarah Bundey
C A Clarke

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