At a meeting of the Editorial Committee held on 2 November 1982 some of the matters discussed would, I thought, be of interest to readers of the Journal.

Delays in publication

This is a perennial problem, but Dr Stephen Lock, Editor of the British Medical Journal, was not concerned that for a bi-monthly journal the average wait was around 9½ months. The Journal had received 167 papers in the year, an increase of 16.7% over the previous 12 months, and there were papers in hand sufficient for the December 1982 and the February and April 1983 issues. Nevertheless, several members of the Committee were concerned at the delay and felt that it might deter potential authors from submitting papers to the Journal. Suggestions were made for cutting the delay, including the abolition of refereeing, eliminating some stages in the printing process, and giving priority to important papers. The Editor noted that this had already been done, but it was agreed that an announcement should be published stating that a mechanism existed for expediting papers and that authors could plead their case if they felt their paper warranted early publication. It was also agreed that the date of acceptance of a paper as well as the date of receipt should be published.

Avoiding fuddy-duddiness

There were some interesting views here. One member thought that we should try and keep up with molecular biology and hoped that papers on this discipline would be forthcoming. Another felt that since the Journal was intended mainly for clinical geneticists we were likely only to get 'cast-offs' on molecular biology. The Editor feels that such polarisation is not inevitable and that review articles on molecular genetics might help to solve the problem.

Review articles

The numbers, though not the quality, here have been disappointing. It was felt that a greater effort should be made to obtain papers from younger authors, and it was agreed to approach heads of clinical genetics units with this in view. An increase of the fee to £75 might make the proposition more attractive. The Editor felt that it would increase the interest in the Journal if some of the views were speculative, for example on maternal inheritance or 'risk factors', and that on the experimental side topics such as the fragile X, aneuploidy, or preventing congenital abnormalities would be very suitable.

Association with Societies

It was generally agreed that the Journal should not be the mouthpiece of either the Clinical Genetics Society or the Association of Clinical Cytogeneticists, though friendly relations with both should be maintained, and the Editor felt that the publication of abstracts of meetings had been a useful exercise. It was agreed that each of the Societies should nominate a member to the Editorial Committee.

Human gene mapping nomenclature

At the meeting in May 1980 a new nomenclature for human gene mapping was discussed, but it was decided not to adopt this. At the meeting on 2 November 1982 the subject was reopened and later Dr Bodmer wrote that his quibbles with the new nomenclature had related to his own pet (HLA) system because of changes which were in contradiction to the then accepted international nomenclature. His criticisms were noted and he now thinks the amended proposals are good and worth following. The Editor therefore thinks that we should accept the new nomenclature.

'Lateral reading'

Some years ago the Editor had suggested that readers should send in extracts of interest from other journals, but this idea had never properly got off the ground. It was now felt that the Editor should be in charge of such a column, which might be designated 'Lateral reading'. He would keep a look out for items himself but would also welcome contributions. The following examples will show how he spent his Christmas.

Cancer and immortality (see Newbold RF, Overell RW, Connell JR. Nature 1982;299:633–5)

Limited lifespan in normal diploid mammalian cells in culture is well recognised, whereas cells derived from malignant tissue often appear to be immortal. Newbold et al have carried out experiments with Syrian hamster cell cultures and the results suggest that normal mammalian cells can be induced to

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escape from their commitment to senescence. This is
effected by exposing them to various mutagenic
carcinogens, though the resulting capacity for
infinite multiplication precedes (and may be necessary
for) malignant transformation. I recall J B S
Haldane’s poem “Cancer is a funny thing”.

PRESENILE CATARACT (see Winder AF. Trans
Various studies have shown that presenile cataract
may be caused by the heterozygous state for galacto-
kinease deficiency, but the above is the first paper, as
far as Dr Sarah Bundey knows, which describes the
risk for offspring of heterozygous mothers. Earlier
observations have confirmed that among patients
with presenile cataracts there is an excess of hetero-
yzogotes for galactokinase and galactose-1-phosphate
uridyl transferase deficiency, over what one would
expect from the population frequencies. It is clearly
important to consider these biochemical states in
persons with presenile cataracts, but particularly so
if the patient is a woman of childbearing years.
It had earlier been observed that mothers of children
with infantile cataracts had a lowered distribution
of galactokinase levels compared to controls and in
1981 specific examples were reported by Winder.
He described four families where mothers hetero-
yzogous for galactokinase deficiency (but only
recognised as such after the births of their affected
children) had children with congenital cataracts.
The size of the risk for this occurring in the offspring
of heterozygous women cannot be judged from
Winder’s paper. However, it is likely that these
cataracts are preventable and so ophthalmologists
should be on the look out for galactokinase hetero-
yzogotes who present either with presenile cataracts
or with a child with congenital cataracts. The
cataracts associated with galactokinase deficiency
are thought to be the result of a direct effect on the
lens of galactose and galactitol, and the cataracts
resulting from deficiency of galactose-1-phosphate
uridyl transferase are also the result of high levels
of galactose-1-phosphate on the lens. However, the
latter substance does not cross the placenta. Women
who are deficient in either of these two enzymes
should be kept on a restricted galactose diet during
any pregnancy.

ERRORS IN THE DIAGNOSIS OF CYSTIC FIBROSIS
(see David TJ, Phillips BM. Lancet 1982;ii:1204–6)
In seven patients seen over 3 years, cystic fibrosis had
been wrongly diagnosed. The initial sweat test was
misleadingly high in only three cases. In one case no
sweat test had been done, and in four, one or more
normal sweat test results were ignored. As a result of
misdiagnosis four children were sent to schools for
the physically handicapped and one man lost his job
in the police. Despite warnings about the limitations
of sweat tests and dangers of diagnosing cystic
fibrosis without typical clinical features, cystic fibrosis
is wrongly diagnosed in substantial numbers of

DUPUYTREN’S CONTRACTURE (see Ling RSM. J
For some years I have been interested in the inher-
tance of this condition but have only recently seen
the paper by Ling, from which there seems no doubt
that the disorder is usually controlled by an auto-
somal dominant gene. In two of Ling’s pedigrees the
proband had parents both of whom were affected,
and therefore their offspring may have been homo-
yzogotes; a point in favour of this is that both pro-
band had severe lesions. I have asked Mr Ling if
he can update the pedigrees as, if homozygosity is
present in the cases be cites, then all the children
should be affected in the next generation. In his
article Ling makes no mention of the disorder
affecting the feet, but in my experience it may do so,
producing ‘lumps’ which I think are thickened
tendon sheaths, though there is no deformity. Lack
of examination of the feet may result in under-
diagnosis in relatives of those affected.

BLADDER CANCER AND MOLECULAR GENETICS
(see Logan J, Cairns J. Nature 1982;300:104–5;
149–52)
In the issue of Nature of 11 November 1982 there are
two papers and a leader on the way in which cells,
and particularly human bladder cells, may become
malignant. In general, carcinogenesis is thought to
be a multi-step process and one of the first stages
may be the activation of a cellular gene whose
function usually has nothing to do with cancer but
with normal embryonic development. Various
factors, for example viral infections, a chance
mutation, or an environmental effect, may convert
the proto- into an oncogene. The important develop-
ment is that the oncogenes can be identified by
certain retroviruses, that is, those whose genome can
exist in both RNA and DNA forms. The oncogene
of one line (EJ) of human bladder carcinoma cells
has now been identified by the retrovirus technique
as differing from the normal by the alteration of one
nucleotide, and this single mutation may well have
been environmentally determined (knowing of the
occupational hazards in bladder cancer). In another
line of human bladder carcinoma (T24) the proto-
oncogene is also activated by a single amino-acid
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substitution. We are therefore getting close to the molecular basis of malignancy. But why not carry out the reverse experiment to test the hypothesis? Carcinoma of the oesophagus is sure to occur in certain persons who suffer from tylosis (Howel Evans et al. Q J Med 1958;27:413). Can one not work backwards to show how these persons differ molecularly from their non-tylotic sibs? Every time I ask this question the reply is: "We are not working on this".


In a letter, the authors raise again the already well-ventilated problem of whether or not people at risk for Huntington's chorea wish to know what the future holds. Of 40 well-informed subjects from affected families, 16 were willing to be tested with levodopa, knowing that there was the risk of transient choreic movements, and a further five wished to know the truth provided there was no risk of transient choreic movement. The authors feel that those at risk for Huntington's chorea are far more receptive to provocative testing than perhaps their physicians or even researchers have realised. This is all very well, but about half the subjects thought exactly the opposite. What do readers feel, and how reliable is the levodopa test?

C A CLARKE