Letters to the Editors

On the New Policy for Reports on Chromosomal Anomalies

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It has been announced (Hum. Genet. 51, I—II, 1979) that in the future, *Human Genetics* will publish only exceptionally chromosomal case reports, even if the standard of cytogenetic analysis is beyond reproach. The reason is that “a stage of scientific development in clinical cytogenetics appears to have been reached where single case reports will only rarely carry enough intrinsic interest to justify separate publication. Only collection of cases of a certain aberration together with careful assessment of phenotypic variability will help to add to our knowledge of human chromosome abnormalities—”. Investigators who observe a case with a rare aberration will be “encouraged to announce their findings in one sentence in a special section of the journal and to ask colleagues whether similar cases had been observed”. Once a sufficient number of identical or similar cases has been collected, they can be studied, compared and published as a joint report.

There have been published many case reports that have described the 10th or 20th case in the literature. Some of them have described the same abnormal karyotype as others except that the extra segment was translocated to say no. 4 instead of no. 5 and have claimed the case to be the first in the literature. It is obvious that these case reports have added minimally, if at all, to our knowledge of human cytogenetics. On the other hand, many important chromosome-abnormality syndromes have been published first as single case or family reports. Examples include trisomies of nos. 13, 18, 8 and 9; partial trisomies of 1q, 3p, 3q, 4q, 5p, and 7q; partial monosomies of 18p, 18q, 4p, and 9p. Isn't it fair to say that these case reports, describing the first cases in the literature, have given a significant impact on human cytogenetics? There still remain to be described many partial trisomies or monosomies and important new syndromes may reasonably be

expected to emerge from them. The indiscriminate application of the joint-report policy may cause gross delay in the publication of case reports, especially when the abnormality is rare or needs sophisticated techniques for its identification.

I understand that in astronomy there is a system of registration of new discoveries. A person who finds what is possibly a new celestial body reports it to a registration center. Many reports may be received from all over the world. The discovery of a new celestial body is accepted as conclusive when it is described by at least three independent observers. It then receives the names of the first three who reported it, e.g. the Brown-Smith-Wilson comet. It is obvious that, once the presence of the new comet is established, additional reports (10th, 15th etc.) will not contribute much unless they describe additional new findings or later changes.

It appears that this system with some modifications is quite applicable in the field of human cytogenetics. Only case reports claiming zero, one or two previous cases (could be 3, 4 or 5) at the time of submission would be considered for publication in *Human Genetics*, and the claim would be checked by editors or reviewers. Any papers rejected because of this policy, could then be published as joint reports following the procedure suggested in the announcement.

This policy would drastically reduce “the volume of cytogenetic articles” in *Human Genetics* without losing any significant contributions. In addition, unnecessary delay would be avoided as far as most reports of “new” cases are concerned. The amount of work for editors and reviewers would also be reduced as most of the case reports currently published would not be acceptable under the new policy.

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