Journal of

MEDICAL GENETICS

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Papers, which should be in triplicate and in the Vancouver style (*Br Med J* 1982;**284**:1766–70), should be sent to the Editor, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR. Papers from the USA can be submitted to the North American Editor, Dr P M Conneally, Department of Medical Genetics, James Whitcomb Riley Hospital for Children RR129, Indiana University Medical Center, Indianapolis, Indiana 46223, USA. Submission of a paper will be held to imply that it contains original work which has not been previously published. The signature of each author is required on the covering letter. Permission to republish must be obtained from the Editor.

Papers should conform to one of the following categories.
Original contributions on clinical or laboratory aspects of medical genetics in man and on related animal studies.
Case reports or family reports with particularly instructive clinical or genetic features: to be no longer than 1000 words, with no more than three figures, one table, and eight references.
Short reports: to be no longer than 500 words with a clinical photograph and partial karyotype, if appropriate, and no more than three references.
Review articles will generally be by invitation, but suggestions from authors wishing to prepare a review article will be welcomed.
Short communications and *Technical notes* will also be considered.
Letters to the Editor in relation to papers and to other relevant topics will be welcomed.

Publication of papers thought to be of special importance may be expedited.

SI units should be used. All contributions should be accompanied by an abstract or structured abstract giving the main results and conclusions. Typescripts should be double spaced with wide margins. One page proof will be sent to the author submitting the paper and alterations on the proof, apart from printer's errors, are not permitted. Reprints may be ordered when the proof is returned.

Figures should be kept to a minimum and should be numbered consecutively in Arabic numerals. Legends should be typed on a separate sheet. Photographs should be on glossy paper and diagrams should be drawn on stout white paper. Photographs of karyotypes do not reproduce well. Chromosomes should be cut out and stuck onto stout paper. Any lettering should be indicated on a separate transparent overlay. Colour printing can be undertaken.

Tables should not be included in the body of the text, but should be typed on separate pages and numbered with Arabic numerals. A legend should be provided.

References should conform precisely to the style current in this Journal. Authors are responsible for the *accuracy* and *completeness* of their references as these will not be checked by the Editorial Office.

Some notes on nomenclature can be found in *J Med Genet* 1991;**28**:72.

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ISSN 0022–2593

Published by British Medical Association, Tavistock Square, London WC1H 9JR, and printed in England by The Devonshire Press Ltd., Torquay

testing was performed. Neither of his parents was heterozygous for △F508 deletion of the cystic fibrosis gene. The third patient, a male born in 1987, had a high IRT activity of 73 μg/l (upper limit of normal range 80 μg/l). He died before sweat testing could be carried out.

The raised IRT levels were probably the result of pancreatic anomalies associated with trisomy 13.[3] In aborted fetuses with trisomy 13, Gosden and Gosden[4] found extensive changes in the pancreas which was larger than in normal fetuses and was intensely pink, probably owing to islands of spleen cells, especially in the tail. The pancreatic tissue was disorganised with blockage or atresia of the main pancreatic duct. Although the pancreatic ducts proximal to the atretic main duct were moderately dilated, they did not show the abnormal content usually seen in the cystic fibrosis pancreas. Although the authors[4] did not measure blood IRT, the histological changes in the pancreas would suggest that the IRT would have been raised.

During the same screening period, 26 infants were born with trisomy 18, all of whom had a normal IRT test. Interestingly, Heeley and Fagan[5] reported two cases of trisomy 18 with a raised level of IRT. In one patient, postmortem histological examination showed focal increases of extra-acinar ducts in the interstitium between the exocrine lobules with some of the abnormal ducts showing inspissated secretions. In the second case, the IRT level had returned to a normal level by 9 days. None of our trisomy 18 patients had raised IRT levels. Our policy of screening between 5 and 8 days may have missed transient rises of IRT.

We consider that our three cases of trisomy 13 did not have cystic fibrosis, but that the positive IRT values may have been the result of associated pancreatic anomalies.[3 4] We would be interested to hear if any other centre with routine IRT screening has had a similar experience.

We thank Mr B Sheridan and Mr R Gamble for their assistance in tracing the IRT results of the patients.

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1 Nevin GB, Nevin NC, Redmond AO. Cystic fibrosis in Northern Ireland. *J Med Genet* 1979;**16**:122–4.
2 Roberts G, Stanfield M, Black A, Redmond AO. Screening for cystic fibrosis: a four year regional experience. *Arch Dis Child* 1988;**63**:1438–43.
3 Schinzel A. *Catalogue of unbalanced chromosome aberrations in man*. Berlin: de Gruyter, 1984:505–10.
4 Gosden CM, Gosden JR. Fetal abnormalities in cystic fibrosis suggest a deficiency of cholecystokinin. *Lancet* 1984;ii:541–6.
5 Heeley AF, Fagan DG. Trisomy 18, cystic fibrosis and blood immunoreactive tryspin. *Lancet* 1984;i:169–70.

NOTICES

Conference on predictive testing for Huntington's disease

There will be a conference on 'Predictive Testing for Huntington's Disease' on Wednesday 13 November 1991 at the King's Fund Centre, London. The Conference will discuss a national protocol and the contribution of genetics, neurology, and psychiatry. Details from: Huntington's Disease Association, 108 Battersea High Street, London SW11 3HP. Tel: 071 223 7000.

International standing committee on human cytogenetic nomenclature

During the 8th International Congress of Human Genetics in Washington, DC on Thursday 10 October 1991 at 6.30 pm (see programme for venue), an open meeting of cytogeneticists will be held. At this meeting a proposed addition to the ISCN nomenclature for the description of abnormal karyotypes in cancer cells will be presented by Felix Mitelman who has chaired a subcommittee working on this proposal. In addition, three new members of the seven member ISCN committee will have to be elected. Nominations of cytogeneticists interested in serving on this committee should be submitted before 1 August 1991. Cytogeneticists who are interested in voting but are unable to attend the International Congress should request voting papers before 1 September 1991. Contact: Dr Uta Francke, Chair, International Standing Committee on Human Cytogenetic Nomenclature, Department of Genetics, Stanford University School of Medicine, Stanford, California 94305–5120, USA. Fax: (415) 725–8112.