better with the *AluI*-Giemsa technique.

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Cystic fibrosis in Bulgaria

In an article published in the journal, Cuppens *et al*¹ described a cystic fibrosis patient who was homozygous for the G542X mutation yet presented with a relatively mild clinical picture.

This mutation, although rarer than $\Delta F508$, seems to occur in all European populations. In the Bulgarian CF patients we are testing it is found at a frequency of 5% of CF alleles. So far we have detected six patients with G542X including one homozygote (table). The clinical course of the disease is invariably severe in our patients. Symptoms of CF had been present from the first months of life and in all cases the diagnosis had been established during the first year. Taking into account the fact that CF is often diagnosed late in this country, early diagnosis suggests a severe course of the disease. At present only one out of the six patients with G542X is still living. Early infant death occurred in four of the families (in two of them a previous affected

Bulgarian cystic fibrosis patients with G542X

Patient No	Mutations	Family and clinical data
33	G542X/G542X	Index patient and affected sister died in infancy
3	G542X/ $\Delta F508$	Index patient and affected brother died in infancy
54	G542X/ $\Delta F508$	Pulmonary involvement, still living
74	G542X/ $\Delta F508$	Meconium ileus, died at 4 days
28	G542X/S549N	Pulmonary involvement, died at 7 years
66	G542X/N1303K	Meconium ileus, died at 10 days

child had also died at a very early age). Two patients had meconium ileus (MI); this has been recorded in about 10% of our CF patients and thus seems to be over-represented in the G542X subgroup (seven MI out of a total of 78 CF patients and two MI out of six with G542X).

Our findings are thus in agreement with the assumption that a stop codon at position 542 results in the synthesis of a functionally inactive protein. On the other hand an increasing number of publications, as well as our own observations, suggest that in a number of patients the actual clinical findings conflict with the molecular evidence. Heterogeneity within groups with the same molecular defect could suggest the existence of additional, perhaps genetic, factors which modify the clinical course of cystic fibrosis.

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Floating Harbor and the good ship Shprintzen

I would like to take issue with the authors of the 'Syndrome of the month' concerning the Floating Har-

bor syndrome,¹ particularly their penultimate patient number 1. This looks to me very much like a case of velocardiofacial (Shprintzen) syndrome on the basis of facial phenotype, especially speech delay, immobile palate, malocclusion, and pulmonary stenosis. Pulmonary stenosis can be associated with the Shprintzen syndrome.^{2,3} The facial dysmorphism appears typical in this photograph: deficient alae, high nasal bridge, almond shaped palpebral fissures, myopathic facies, and maxillary and mandibular hypoplasia. This patient does not have a large mouth. Immobile palate is of particular interest and I would suggest that this patient be reviewed as regards submucous cleft of the palate. Even if it is not obviously present, it may well be at operation. Videofluoroscopy of the palate would be of some assistance in this regard. These patients are uniformly unresponsive to speech therapy and respond only to palate lengthening or pharyngoplasty or both.

It is clear that if there is some considerable doubt about the first patient the other reported patients need to be reviewed. The details supplied were not adequate enough to determine if other syndromes or small, odd-looking children have been lumped together under this enticing name. In particular, actual height centiles would be of interest. Patients with the velocardiofacial (Shprintzen) syndrome are small, but not several standard deviations below the mean.

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BOOK REVIEW

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.

IVF and Justice: Moral, Social and Legal Issues Related to In Vitro Fertilisation. Teresa Iglesias. (Pp 175; £9.75.) London: The Linacre Centre for Health Care Ethics. 1991.

Intrinsic to the human condition is our susceptibility to outside influence. Social acceptability of a practice tends to alter our moral outlook to accommodate that practice. These accommodations in social conscience do not, however, question the basic correctness or otherwise of that practice. In vitro fertilisation forms a case in point. It marked a quantum scientific breakthrough in the treatment of infertility. Latterly, the unwanted side products of 'surplus' embryos have become legitimate subjects of scientific research up to the 14th day of their existence. Surrogate motherhood, the 'ownership' of children resulting from surrogate pregnancies

and the 'principle of anonymity' governing artificial insemination by donor are all issues with which society has had to grapple in consequence of the technological breakthrough which IVF represented. That these are now accepted practices may have altered their moral acceptability to many people, including the medical fraternity, but acceptability and the moral correctness of these practices are not necessarily synonymous.

Philosopher and ethicist, Teresa Iglesias, a graduate of universities in Madrid, Dublin, and Oxford, examines these complex issues, and a host of others, in this book. These issues are judged on their morality or lack of same rather than by the social acceptability barometer. The basic standpoint of this examination is Christian rather than denominational.

The book takes the form of seven chapters, each of which has been presented as a paper examining various aspects of IVF elsewhere. Now they are presented together in book form to offer a comprehensive discussion of the subject as a whole. Topics discussed include the nature of the human embryo, death and the beginning of life, and the claims of the embryo to be recognised as an individual. These topics are examined in light of the practice and consequences of IVF. The style of these chapters is somewhat irksome, each comprising a numbered set of arguments, sometimes related, sometimes not. Thus the style is reminiscent of the school-boy debater, complete with striped blazer, college cap, and tie about to enter the representative fray!

Yet, this is a weighty book dealing with weighty matters and subjects all too easily dismissed by those of us blinded by scientific success to the implications in terms of right and wrong. The legislative position of the human embryo historically is examined. The conclusion from this investigation is that the embryo has always occupied a position of 'human entity' in British law. The recent legislation allowing experimentation on human embryos is inconsistent with this historical position and with

the precepts of basic justice, according to the arguments contained herein. The status of IVF generated embryos has been changed from that of 'human entity' to disposable asset. By so doing the implication is that IVF generated embryos are 'lesser forms of humanity'. The social and legal justification for this is the greater good and benefits which accrue to others, that is, 'consequentialism', whereby the essentially immoral act of generating embryos by IVF is justified on the basis of successfully treating infertility, providing scientific research material of enormous interest, etc. Nevertheless, the pursuit of individual freedom and choice for infertile couples may have led, inadvertently, to the denial of basic human rights to the IVF generated embryos.

Fundamentally, this book asks of IVF whether the many ends justify the means. From every one of the 175 pages the answer of 'no' is screamed. Many will dismiss it as 'Catholic clap-trap' but the more reflective will find much food for thought between these covers, even if they disagree with some of the basic precepts and the ultimate conclusions.

WILLIAM REARDON

NOTICE

Journal of Genetic Counseling: call for papers

The first issue of the *Journal of Genetic Counseling*, the journal of the National Society of Genetic Counselors, will be published in early 1992. Manuscripts addressing any aspect of counselling in genetics are invited. Manuscripts and requests for additional information should be addressed to the Editor Deborah L. Eunpu, Developmental Medicine and Genetics, Albert Einstein Medical Center, 5501 Old York Road, Philadelphia, PA 19141-3098, USA. Tel: (215) 456-8706.