acromegaly. Is the lipid abnormality caused directly by the high levels of growth hormone, or is this due to the well-known disturbance in insulin metabolism?

DR. TULLOCH: Our group of acromegalics was selected for study to exclude those with an abnormal glucose tolerance test. There is little information about how metabolic changes secondary to growth hormone could be differentiated from the associated plasma insulin increase. We once studied a single case of pan-hypopituitarism due to a craniopharyngioma, and recorded an increase of fasting plasma triglycerides from 70 to 120 mg/100 ml after six days' treatment with human growth hormone (10 mg/day).

PROF. RUSSELL FRASER: Well, evidently we still do not know the mechanism of this heart disease which afflicts a high proportion of acromegalics. This problem remains for future study.

This conference was recorded and edited by Dr. G. F. Joplin.

APPOINTMENTS OF SPEAKERS

(1) Dr. G. F. Joplin, Senior Lecturer in Clinical Endocrinology and Consultant Physician, Royal Postgraduate Medical School, and Hammersmith Hospital.

(2) Dr. F. H. Doyle, Reader in Diagnostic Radiology, Royal Postgraduate Medical School.

(3) Dr. Celia M. Oakley, Consultant Cardiologist, Royal Postgraduate Medical School, and Hammersmith Hospital.

(4) Professor Russell Fraser, Professor of Clinical Endocrinology and Consultant Physician, Royal Postgraduate Medical School, and Hammersmith Hospital.

(5) Dr. P. D. Lewis, Lecturer in Histopathology, Consultant Physician and Histopathologist, Royal Postgraduate Medical School, and Hammersmith Hospital.

(6) Dr. C. A. Pallis, Reader in Neurology, Royal Postgraduate Medical School.

(7) Dr. D. J. Evans, Senior Lecturer and Consultant Histopathologist, Royal Postgraduate Medical School and Hammersmith Hospital.

(8) Dr. B. R. Tulloch, Wellcome Senior Clinical Research Fellow and Honorary Lecturer in Clinical Endocrinology, Royal Postgraduate Medical School.

(9) Dr. B. Pimstone, Visiting Colleague (endocrinology).

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Contemporary Themes

Social Effects of Genetic Counselling

ALAN E. H. EMERY, MURIEL S. WATT, ENID CLACK

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Summary

In a follow-up study of 104 subjects referred for genetic counselling between 1965 and 1969 all were at risk of having children with a variety of serious genetic disorders. Most subjects were in social classes III and IV, were married, in their late 20s, and already had an affected child. Sixty-three per cent. were referred by hospital consultants, 27% by their general practitioners, and 10% were self-referrals. All of those counselled appeared to have appreciated the genetic implications, although four overestimated the risks and 11 underestimated the risks.

Of those at high risk (greater than 1 in 10) of having an affected child 10 out of 55 couples "planned" further pregnancies despite the risks. In two this was because they had been unable to adopt a child, in four because they had no living children and the disorders in question usually resulted in stillbirth or death in infancy so that the "burden" of an affected child would be of relatively short duration, and one mother had had antenatal diagnosis and selective abortion. Most of the couples in the low-risk group (less than 1 in 20) were reassured and planned further pregnancies.

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Introduction

An increasing proportion of morbidity and mortality, particularly in childhood, can be attributed to genetic factors. For example, in a recent survey in Newcastle upon Tyne, no less than 42% of childhood deaths in hospital could be attributed to disorders which were largely or even entirely genetically determined.[1] The prevention of such disorders lies within the province of genetic counselling whereby the risks of recurrence are explained to prospective parents. If the risks are high and the disorder in question is a serious one the parents may decide to have no further children and rely on contraception or one of the parents being sterilized. More recently antenatal diagnosis with selective abortion of affected fetuses has become possible in chromosomal and certain metabolic disorders. A follow-up study from this department of women referred for genetic counselling in families with Duchenne type muscular dystrophy showed that some women at high

risk of having an affected child appeared to be undeterred by the risks,[2] yet this is a serious disease for which at present there is no treatment. In view of these findings it was decided to extend this study to other persons seen in the genetic clinic who were considered at the time to be at risk of having a child with a serious genetic disorder.

Subjects and Methods

The families in the present study came from the Manchester and Edinburgh regions and were seen during the four-year period April 1965 to March 1969. Only persons referred *specifically* for genetic counselling and who were at risk of having children with a *serious* genetic disorder were included. Those with disorders which were considered not serious or not genetic were excluded, as were subjects who were less than 18 or over 39 years of age at the time of counselling. Families with Duchenne type muscular dystrophy were also excluded, as this material has been reported elsewhere.[2]

One hundred and sixteen subjects were chosen for study; however, in spite of exhaustive inquiries and a search through hospital files and other records it was not possible to trace 10 of these, and two others were unwilling to co-operate in the study.

The numbers of subjects counselled according to the modes of inheritance of the genetic disorders involved are summarized in table I. Risks of recurrence were based on genetic principles and arbitrarily considered "high" if greater than 1 in 10, "medium" if 1 in 10 to 1 in 20, and "low" if less than 1 in 20. The decision whether to accept the risk of having an affected child was left to the subject concerned, though in some cases where the risk was high this was emphasized. More recently the possibilities of antenatal diagnosis and selective abortion have been offered in appropriate situations.

TABLE I—*Modes of Inheritance of Genetic Disorders in Counselled Subjects*

Inheritance	No. of Subjects	Age when Counselled (Years)					
		18-	21-	24-	28-	32-	36-40
Unifactorial:							
Autosomal dominant	20	2	3	5	5	2	3
Autosomal recessive	30	3	4	8	7	7	1
X-linked recessive	8	—	1	3	1	—	3
Multifactorial	38	1	5	12	12	4	4
Chromosomal	8	—	3	1	3	1	—
Total	104	6	16	29	28	14	11

Results

All social classes were represented, with most people in classes III and IV. Most of those who sought advice were aged 24-31 years (table I). Ninety-five were married and nine were single at the time of counselling. Sixty-three per cent. were referred to the clinic by hospital consultants, 27% by their general practitioners, and 10% were self-referrals.

The majority who attended the clinic were healthy persons who either had an affected child or had a family history of a genetic abnormality and were concerned with the risk to future children. All of those counselled appeared to have appreciated the genetic implications, although four overestimated the risks and 11 underestimated the risks (table II). The mode of inheritance was not always clearly understood, particularly in conditions believed to be of multifactorial origin. Subsequent discussions at follow-up proved very helpful in these cases. In fact, many subjects said that they had felt confused and somewhat overwhelmed by the information given at genetic counselling and would have valued opportunities for further discussions.

The effects of genetic counselling on family planning and marital status are shown in table III. In the high-risk group

TABLE II—*Comprehension of Genetic Counselling by Subjects (Married or Single) Regarding Risk of having an Affected Child*

	Affected Subjects			Healthy Subjects				
	AD	AR	MF	AD	AR	XR	MF	CHR
Completely understood	3	2	3	15	25	6	28	7
Partly understood:								
Overestimated risk	0	0	0	2	0	0	2	0
Underestimated risk	0	0	0	0	3	2	5	1

AD = Autosomal dominant. AR = Autosomal recessive. XR = X-linked recessive. MF = Multifactorial. CHR = Chromosomal.

TABLE III—*Marital Status and Family Planning Subsequent to Genetic Counselling*

Risk	Single		Married			Marital Breakdown
	Subsequently Married	Remained Single	Avoided Pregnancy: Successfully	Avoided Pregnancy: Unsuccessfully	Planned Pregnancy	
High	2	3	41	4	10	2
Medium	0	1	4	0	10	0
Low	2	1	12	1	17	2
Total	4	5	57	5	37	4

TABLE IV—*Families at High Risk who Planned Further Pregnancies Subsequent to Genetic Counselling*

Case No.	Disorder	Genetics	Family History	Outcome
1	Choanal atresia	AR	2 Affected children*	1 Normal child
2	Albinism (with grossly defective vision)	AR	1 Affected child	1 Normal child
3	Ichthyosiform erythroderma	AR	1 Affected child*	2 Normal children
4	Spina bifida, anencephaly	MF	2 Affected children*	1 Normal child
5	Spina bifida	MF	2 Affected children*	2 Affected children
6	Spina bifida	MF	1 Affected child and mother's brother affected*	1 Normal child
7	Becker muscular dystrophy	XR	2 Affected maternal uncles. Mother had raised serum creatine kinase	Normal son and daughter
8	Osteogenesis imperfecta	AD	Father affected	1 Affected child
9	Peroneal muscular atrophy	AD	Father and other relatives affected	2 Normal? children
10	Werdnig-Hoffmann disease	AR	1 Affected child*	1 Normal child

*Affected individual either stillborn or died in infancy.
A key to the abbreviations is given in table II.

five were unmarried at the time of counselling and three remained single, though apparently not because of the genetic risks. One woman underwent tubal ligation before marriage. Of those at high risk of having a child with a serious genetic disorder 10 *planned* pregnancies despite the risks involved (table III). The outcome in these pregnancies is summarized in table IV. In case 8, which resulted in an affected child, the marriage subsequently broke down. In two cases the parents planned pregnancies despite the risks involved because they had been unable to adopt a child. In four other cases the parents planned pregnancies because they had no living children and considered the "burden" to the family of an affected child would be of relatively short duration because the disorders in question usually resulted in stillbirth or death in infancy. One woman who was a carrier of sex-linked Becker type muscular dystrophy had a selective abortion of a male fetus. In 45 cases the parents had intended to avoid further pregnancies because of the genetic risks.

In most cases the responsibility for family planning was left to the wife (table V), tubal ligation or oral contraception being preferred. More recently, however, more fathers have considered vasectomy because of its comparative simplicity and reliability. In the four cases where contraceptive measures failed all had a therapeutic abortion. One mother conceived

TABLE V—*Contraceptive Measures Adopted by Married Couples Subsequent to Genetic Counselling*

Risk	Total No.	No Contraception	Contraception					
			Oral	T.L.	Vasectomy	I.U.D.	S./D.	Rhythm
High ..	55	10	16	14	3	3	6	3
Medium	14	10	3	0	0	0	0	1
Low ..	30	17	6	2	0	2	1	2
Total	99	37	25	16	3	5	7	6

T.L. = Tubal ligation. I.U.D. = Intrauterine device. S./D. = Sheath or diaphragm.

subsequent to tubal ligation and had a hysterectomy at the time of termination. One relied on an intrauterine device and two relied on the sheath; of the latter one subsequently chose oral contraception and the other tubal ligation.

In the medium-risk group 10 couples planned further pregnancies, resulting in 10 unaffected children, one spontaneous abortion, and a child who was later diagnosed as having cerebral palsy. In the low-risk group 17 couples were reassured and planned further pregnancies, resulting in 15 unaffected children and one spontaneous abortion; one mother was pregnant at the time of follow-up. Twelve couples had no further children subsequent to genetic counselling; six felt reassured and planned to have children in the future, three considered that they had sufficient children, two were divorced, and one couple had a severely handicapped child with spina bifida, and for this reason avoided further pregnancies. One mother who conceived had a therapeutic abortion because of her fear of having another child with Down's syndrome, though neither she nor her husband was a translocation carrier.

There were four cases of marital breakdown resulting in separation and divorce, and much of the difficulties were attributed to sexual problems consequent on the fear of having an affected child even though two of these couples were in the low-risk group.

The need for help and support from social welfare and voluntary services was apparent in many families adjusting to the problems of chronic disability in either a child or parent. There was often considerable strain within family units, with feelings of guilt where a child was involved and frustration consequent on increasing dependency in the case of affected adults. The counselling service was greatly appreciated by most of the families seen, and many expressed the need for further discussions and counselling for themselves or other family members. The genetic counselling clinic appeared to have provided subjects not only with genetic advice and explanations of the nature of particular disorders but also with a forum in which they could discuss their anxieties, fears, and feelings of guilt.

Discussion

The follow-up of persons who have had genetic counselling is valuable for several reasons.[3] Firstly, it provides a chance for the counsellor to assess whether the information he has given has been properly understood and interpreted. Secondly, it provides an opportunity to reassure parents that active research into the cause and possible treatment of a particular disease is being carried out, to put families in contact with various welfare agencies, and to discuss recent developments such as selective abortion. Thirdly, it allows the geneticist to assess whether other members of the family are at risk of having affected children and require genetic advice. Concern with the latter problem led this department to develop a computerized genetic register system referred to by the acronym "RAPID"—Register for the Ascertainment and Prevention of Inherited Disease.[4,5]

Unfortunately few previous studies have been concerned with the possible social effects of genetic counselling. In the study by Carter *et al.*[6] more couples came from the upper social classes and all the families wished for advice, which may explain why so many acted "responsibly." Leonard, *et al.*[7] reported their observations on the effects of genetic counselling in an unselected group of families with children with fibrocystic disease, phenylketonuria, and Down's syndrome. The apparent comprehension of genetic advice is an important consideration. Leonard *et al.*[7] found that 19 out of 61 couples completely failed to understand the information provided and that eight understood only imperfectly. It would not be fair to indict genetic counselling in general on this evidence, for in this study advice was provided by a number of agencies, presumably not all with the same effectiveness. In the present study of 104 subjects all appeared to have appreciated the genetic implications, though in 15 the risks were apparently not remembered precisely. This may, however, have been more the result of wishful thinking than a lack of comprehension, since in 11 of these cases the risks were *under* estimated. Rather disturbing findings in the present study were that a number of couples at high risk of having a child with a serious genetic disorder were undeterred from having further children, and in four out of 45 of those in the high-risk group who had intended to avoid further pregnancies contraceptive measures failed. Failed contraception and serious marital disharmony resulting from the fear of having an affected child suggests that expert contraceptive as well as genetic advice is needed in these cases.

The comprehension and appreciation of genetic risks and their implications might be improved in several ways, many of which were suggested to us by parents during the course of this study. Firstly, a second clinic visit seems to be essential, particularly when the first visit may have been largely devoted to establishing a precise diagnosis. Perhaps a written summary of the facts would also be helpful as well as a follow-up discussion in appropriate cases some 6 to 12 months later. Secondly, genetic advice should not be given too early after the birth of an affected child or the establishment of the diagnosis of a genetic disorder. The emotional shock may well interfere with the comprehension and acceptance of advice. Thirdly, the amount and nature of the information imparted should obviously be geared to the educational background of the individual. We think that the emphasis should be on explaining the nature and prognosis of the disease in question (since this appears to influence potential parents as much as the genetic risks) and genetic mechanisms explained only in the very simplest of terms. Principles should be emphasized rather than mathematical probabilities. In our experience the greatest difficulty in genetic counselling is the lack of biological knowledge of most couples seen in the clinic. Here the responsibility seems to lie with educationalists. It seems reasonable nowadays that discussions of the biology of reproduction and sex education in schools should be broadened to include the basic principles of genetics and the causes of congenital abnormalities. Only then will a really meaningful rapport between the medical geneticist and most couples be possible.

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