

## **Introduction**

Throughout the year genetic issues have continued to occupy centre stage in the media and politics. Genetics seems to have all the elements necessary to make a good story. It is “big science” with governments, universities and multi-national corporations all actively involved. The new knowledge revealed by research seems to hold out either great promise or threat of disaster, leading, too often, to a kind of feeding frenzy in the media which often seems to bear little or no relationship to the reality of the science on which these stories have been based.

The impact of this attention, hype and hyperbole on the circumstances of families living with the day to day reality of a serious genetic disorder affecting themselves, their children or those they love is often overlooked. Yet it is these people who have the most to hope for from developments in science and medicine arising from human genetic research and who have most to lose if this new knowledge is not put into effect, or if it is abused.

For the trustees, staff and members of the Genetic Interest Group this year has been focussed on making sure that the needs of and values of families affected by serious genetic disorders are right at the heart of the debate about scientific progress, its potential benefits and the ethical, legal or social issues that are raised by continuing new knowledge.

### **Working with Government.**

During the summer of 2000 the Government published its long awaited response to the report of the Chief Medical Officer's expert enquiry into the potential uses of embryonic stem cells in research and for the development of treatments for serious diseases of adulthood. In accepting all the recommendations of the report the Government announced that there would be a free vote when Regulations amending the Human Fertilization and Embryology Act were introduced.

Working alongside other charities, including the Association of Medical Research Charities and the Parkinson's Disease Society and with the active support of many of our member groups, we set out to persuade MPs and the Lords about the potential benefits this research might yield for those living with the consequences of devastating disorders. We recognise the sincerely held beliefs of those opposed to this work. However after canvassing and consulting the views of our members and scientific experts, we argued that the potential for good was such that this work should actively be encouraged. We were delighted when, as a result of intensive activity by GIG and others, both Houses of Parliament voted in favour of amending the HFE Act and allowing this work to proceed by an overwhelming majority.

As well as looking to the brighter future that research holds out, we have been closely involved with improving the services offered now by the NHS for those who need them. GIG representatives were members of a working party of the Department of Health charged with looking at the structure of genetic laboratory services and the

resources needed for them to do their job properly. A major result has been the recognition of the need to create a network of labs working together and sharing out testing for rare genetic disorders in a logical and rational way. This plan has been adopted by the Department and the practical arrangements are now being worked out by the national Genetics Commissioning Advisory Group on which GIG has a seat and which reports directly to Sir John Pattison, the Director of R & D for the NHS.

We have also played an active part in the Public Health Genetics Network, which focuses on the broad implication of genetic advances for the NHS and in the work other committees, including the Joint Committee on Medical Genetics of the Royal College of Physicians, the Royal College of Pathologists and the British Society of Human Genetics - working in partnership with the people who determine the shape of services provided by the NHS.

In all these settings GIG has often been the sole voice putting forward the views of individuals and families who know for themselves what life is like with a genetic disorder. We depend on the active engagement of our members and their willingness to commit their time, energy and enthusiasm to making sure that GIG really can speak on behalf of its members with confidence and authority.

## **The Human Genetics Commission**

As the major source of policy advice to Ministers on issues relating to genetics and human health we have devoted a lot of attention to cooperating with the Human Genetics Commission. Many of GIG's members were unhappy when the membership of the commission was originally announced that it did not include a broader range of members with direct experience of the spectrum of genetic disorders affecting themselves and in their families. Following pressure on the minister and the HGC discussions have been going on as to ways in which the Commission can have input from those who have experience of this diversity. Although progress has been slower than we would have wished, it has been agreed that a consultative forum will be established that can act as a sounding board for commissioners wishing to test their views against the reality of people's experience. We are collaborating closely with the chair of the HGC's public consultation sub-committee and the secretariat in the Department of Health to make this happen.

During the year the Human Genetics Commission published a wide ranging consultation document, covering issues as diverse as the forensic use of DNA, genetics and insurance and the creation of large scale databases. GIG's response was generated after an extensive consultation programme with its member groups and formed a substantial reaction to the issues raised by the Commission. It was notable for the emphasis that arose from our members of the need to be realistic in our appreciation of the power and the potential impact of genetics and not to legitimize unrealistic anxieties by giving them undue weight.

The HGC's response to their consultation will be published in the Autumn of 2001. We will be looking closely at this when it comes.

## **Genetics and Insurance**

The use of genetic information by insurance companies has been a hot topic this year. In our view too much emphasis has been placed on genetic information revealed by DNA tests at the expense of other forms of predictive information such as family history. GIG has campaigned hard to broaden the debate so that the relevance of all types of genetic information can be looked at, not just the results of DNA tests. We have also argued for the need for proper social welfare benefits for all those affected by genetic disorders as a must, rather than relying on private insurance as a means of providing for individual wealth and well being when genetic disease occurs.

As a member of the UK Forum of Genetics and Insurance and the Association of British Insurers Genetics Committee we have also engaged in an educational role with underwriters and geneticists to create better mutual understanding of the way in which both of these groups go about their business.

## **GIG and its members.**

The "Consortium" project, funded by the National Lottery Charities Board (now the Community Fund) concluded at the end of the year. This provided substantial benefits for those organisations which took part. One of the most exciting outcomes was the establishment of "Exsight" as an independent charity in the West of England. This provides social opportunities for people with genetic forms of blindness. Once this was up and running GIG stepped back, as the

new charity had reached the point where it was able to develop under its own momentum.

We also held a series of training workshops for small groups on topics of common concern. These were made possible by a grant from the Baring Foundation. Typical of these was one for support groups for people with intersex conditions. This provided training for volunteers working telephone helplines for these groups, giving them the skill and confidence to deal with sensitive and potentially stressful situations.

GIG has been asked to provide speakers to members' conferences and organise workshops to facilitate sharing experiences thus enabling everyone to work better and more effectively. These have been very well received and we would do more if our resources allowed.

### **GIG and individuals**

Every year more than 600 people contact the GIG office asking for information and support. Most of these are individuals or families, often newly diagnosed, seeking someone to explain their situation, able to help them understand what the implications of their condition might be.

Linking these people with relevant support groups or expert professionals is a central element of GIG's work, helping to cut a way through conflicting advice and confusing information to sources of practical help and guidance from those who really know.

In addition we are often approached by professionals seeking

information on behalf of their patients or clients, so improving their own effectiveness and making their input more relevant.

To-day's students are tomorrow's professionals. For many, the excitement of genetics and the potential it holds out is a source of inspiration. GIG is happy to respond to requests for help with projects and dissertations. This year we have been able to help students working on projects for GCSE, A-level, undergraduate and post-graduate qualifications.

### **The Media**

As always, media attention to genetic issues has made demands on GIG's staff and trustees. Often behind the scenes briefing has helped reporters and media researchers realise that what at first sight may have seemed like a major story is in fact much less significant. Taking the hype out of genetics is important in preventing false hopes from being built up or fuelling alarm when it is not justified. GIG spokespeople have appeared regularly in the national press and on TV and Radio. In addition we have published articles in journals and magazines including the British Medical Journal, Nature Biotechnology and The Biochemist to name but three.

Always keen to stress the importance of relating science to the lives of those with serious genetic diseases we are pleased to have maintained our position with the media as an authoritative source of comment and opinion on the genetic issues of the day and have acted as a conduit for families who are willing to talk to the media about their own conditions..

## **Educating the Professional Community**

Our members report that, for many families, a key problem is getting to see someone who knows about their condition and can offer them real help and support. For GP's, nurses and other medical professionals who are not specialist geneticists, knowing about each of the 5,000 + rare genetic disorders in detail is clearly an impossibility. Recognising that a condition may be genetic is not an impossibility and GIG's programmes of conferences and workshops for family doctors, NHS managers, nurses and other health care workers has continued to raise awareness of families' needs and the opportunities for the NHS to respond to them. GIG's educational programme for primary care workers and for family doctors is one of the largest in the NHS. Even so it barely scratches the surface and much more could be done if more resources were to be made available. We are particularly grateful to the PPP Healthcare Medical Trust for its support with this aspect of our work.

## **Europe**

This year saw the implementation of the long awaited Orphan Medical Products regulations. These have opened up the floodgates for applications from companies wanting to develop products for diagnosing and treating rare genetic diseases. In the first few months the Orphan Medicinal Products Committee at the European Agency for the Evaluation of Medicinal Products has recommended over 70 products for designation. With a seat on the committee, we can see how this measure has provided a real boost for researchers in the field. GIG is also lobbying hard to make sure that, once the research delivers new medicines the NHS is prepared to purchase them for all

who need them. This will be ongoing work.

During the year the European Parliament set up a special temporary committee on genetics. We have made regular inputs to this committee always trying to bring the focus of its members' deliberations on the reality of the situation as it affects our members and their families, emphasising the need to regulate based on sound science and real needs, not speculation or public clamour alone. The committee will report to the European Parliament at the end of 2001. The recommendations the Parliament will make as a result of this report will be keenly scrutinized.

## **Reaching other audiences**

GIG speakers have addressed a wide range of lay and professional audiences in the UK and elsewhere in Europe on a wide range of diverse topics. We have addressed local meetings for our member groups (for example the UK Thalassaemia Society) and major international conferences of the Drug Information Association, the Swedish Presidency of the EU, the International Congress on Human Genetics and many others. GIG staff and trustees have also participated in many other workshops and seminars, often as the sole representative of individuals and families amongst professionals from medical, legal, academic and other disciplines, flying the flag for the family and making sure that what comes out resonates with their needs and experiences.

## Fundraising

GIG draws its funds from a wide range of different sources. We are grateful to all our supporters (listed alongside), without whom we would be unable to continue our work as an advocate and campaigner on behalf of all our member groups and the individuals and families they support. Preserving our independence is vital to our continued success. GIG accepts no donations that ask us to argue for positions not in accordance with the needs and interests of those living with genetic disorders.

All our sponsors recognise this and we have been able to enjoy support from them without pressure or influence being brought to bear on our policy or practice, even when this involved us taking critical stances with which they might have felt uncomfortable. We appreciate the strength and maturity of the relationship we enjoy with our funders that makes this possible.

ABI	2,000
ABPI	5,000
AstraZeneca plc	2,000
Barings Foundation	27,950
BioIndustries Association	2,000
European Foundation for the Advancement of Medicine	4,970
Englehorn Foundation for Rare Disorders	4,988
Friends Provident	3,000
Medical Research Council	15,500
Mercers Trust	1,000
National Lotteries Charities Board	44,050
Novartis	5,000
Nycomed Amersham	1,500
Pfizer	1,000
Prudential	1,000
Serono International	1,336
SmithKline Beecham	25,000
Wellcome Trust	18,150
Other donations	820

**Income and Expenditure**

	<b>2001</b>	<b>2000</b>
<b>Income</b>	<b>£</b>	<b>£</b>
Membership	8,621	8,128
Grants and Donations	166,264	200,289
Meeting Receipts	5,023	2,880
Sundry Income	52	531
Bank Interest	7,257	6,397
<b>Total</b>	<b>187,217</b>	<b>218,225</b>
<b>Expenditure</b>		
Purposes of the charity	150,747	171,337
Fundraising	12,398	3,411
Management and Administration	32,814	33,371
<b>Total</b>	<b>195,959</b>	<b>208,119</b>
<b>Net Surplus/(Deficit)</b>	<b>(8,742)</b>	<b>10,106</b>

The figures reflect a difficult financial climate. GIG has had to work hard to maintain its financial position over the year. Competition from larger charities has squeezed funds, whilst the cost of keeping all our members and supporters informed (a large element of our central administration costs) has remained constant. As an Alliance this aspect of our work is central to our success. Without the involvement and support of members we could not fulfill our charitable purpose, so this aspect of our expenditure will continue to be significant in the future.

**TRUSTEES**

John Dart  
Joanie Dimavicius (Chair)  
Chris Friend  
John Gluckstein (Honorary Treasurer)  
Dee Heaps (Honorary Secretary)  
Ann Hunt  
Charles Kingdom (resigned February 2001)  
Susan Lewis  
Maggie Ponder (Vice Chair)  
Philip Webb

**COOPTees**

Dr Martin Fielden  
Anthony King (resigned July 2001)  
Debbie Kirklin  
Ann Philips

**OBSERVERS**

Dr Shirley Hodgson, FRCP, DM

**ADVISORS**

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Professor Michael Connor, MD, FRCP, DSc  
Professor Timothy M Cox, MD, FRCP, FmedSci  
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Professor Marcus Pembrey, MD, FmedSci, FRCP, FRCPCH, FRCOG  
Professor Sir D Weatherall, FRS, FRCP, FRCPath

## STAFF

### Director

Alastair Kent

### The GIG Team

Sophie Brown (National Co-ordinator) until April 2001

John Gillott (Policy Officer)

Saskia Ottignon (Administrator)

**Registered Charity No 803424**

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## GIG MEMBER GROUPS

Aarskog Syndrome Support Group (UK)

Adrenal Hyperplasia Network

Albinism Fellowship

Alpha 1 Support UK

Alstrom Syndrome UK

Androgen Insensitivity Syndrome Support Group

Anorchidism Support Group (ASG)

ARC

Assert

Association for Glycogen Storage Disease (UK)

Ataxia

Ataxia - Telangiectasia Society

ATRX Support Group

Adrenoleukodystrophy Family Support Trust

Bannayan-Zonana Support Group

Batten Disease Family Association

Beckwith-Wiedmann Support Group

BEHCETS Organisation

The British Porphyria Association

Cancer Research Campaign

Candid

Cardiomyopathy Association  
CGD Research Trust  
Children's Mitochondrial Disease Network  
CLAPA Reading  
CLIMB  
CMT International UK  
Cohen Syndrome Support Group  
Congenital Adrenal Hyperplasia Support Group  
Craniofacial Support Group  
The Cogent Trust  
CDLS Foundation  
Cri Du Chat Syndrome Group  
Dariers Disease Support Group  
DEBRA  
Diabetes UK  
Down's Heart Group  
Dyskeratosis Congenita Society  
The British Ectodermal Dysplasia Support Group  
Ehlers-Danlos Support Group  
Euro-Ataxia  
FAB UK  
The Fragile X Society

FSP Support Group  
Galactosaemia Support Group  
Gauchers Association  
Gorlin Syndrome Group  
Gwent Huntington's Disease Association Support Group  
Haemophilia Society  
Heart Haven  
Haemochromatosis Society  
Huntington's Disease Association (Colchester Branch)  
Huntington's Disease Association  
Hypomelanosis of ITO Support Network  
The International Autistic Research Organisation  
IGA  
The Jennifer Trust for Spinal Muscular Atrophy  
Klinefelter Organisation  
K S Link  
Laurence Moon Bardet Biedl Society  
London and Home Counties Prostate Cancer Support Association  
Long QT Family Support Group  
Marfan Association  
Motor Neurone Disease Association  
Myotonic Dystrophy Support Group

Society for Mucopolysaccharide Diseases  
Muscular Dystrophy Campaign  
Muscular Dystrophy Support Group  
Nail-Patella Contact Group  
NARA  
Narcolepsy Association (UK),  
NASPCS  
Nystagmus Network  
OSCAR  
Pemphigus Vulgaris Network  
PIA  
PKD Support Group  
Prader Willi Syndrome Association  
Primary Cilary Dyskinesia Family Support Group  
Prostate Cancer Support Association  
The PSP Association  
PXE International  
The Neurofibromatosis Association  
Restricted Growth Association  
British Retinitis Pigmentosa Society  
The Retinoblastoma Society  
Rett Syndrome Association UK

Scottish Huntington's Association  
Seriously Ill for Medical Research (SIMR)  
Shwachman Diamond Support  
East London Branch Sickle Cell Society  
National Sickle Cell Programme  
SOFT UK  
STEPS - The National Association for Children with Lower Limb  
Abnormalities  
Stickler Syndrome Support Group  
Sussex ADHD Support Group  
Syndromes Without a Name  
Talking Health  
Tay Sachs and Allied Diseases Ass  
Telangiectasia Self Help Group  
UK Thalassaemia Society  
Tuberous Sclerosis Association  
Turner Syndrome Support Society (UK)  
UKAN  
Unique  
Williams Syndrome Foundation Limited  
Worster Drought Syndrome Support Group  
XLH Network  
X P Supprt Group

