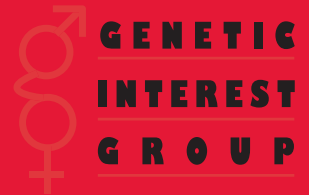




Genetic Interest Group
Annual Report 2001-2002

"Achieving Partnership"



Welcome from the Chairman

This is my first year as chairman so let me introduce myself. I came to the GIG board from the Neurofibromatosis Association where I am a trustee. I have a background in nursing leading through working for a molecular genetics research group and on to where I am now, which is researching on the psychological, social and practical implications of various aspects of genetics and medicine. I have been involved with the Neurofibromatosis Association for all of its 20 years.

I think this year is very well summed up by the words "achieving partnership". The very small team (only 4) of dedicated people who work for us have achieved an enormous amount this year by making active efforts to partner others. These are outlined in detail below but I would particularly like to emphasise GIG's involvement with the Genetic Knowledge Parks, the work with people from ethnic minorities and our continuing effort to break down barriers surrounding medical genetic research. Partnership with our member groups is essential and we wish to be able to involve all of our members more. This is why we undertook the consultation exercise this year. Building on that work we hope that the forthcoming year will see an ever-stronger partnership with them all.

I would like to thank the team - Alastair Kent, John Gillott, Pritti Mehta and Saskia Ottignon for all their hard work through out the year and also for the support I have had from the trustees. I want to thank Joanie Dimavicius, my predecessor in the chair, who has been of special help and has guided GIG so well in the past years.

As always we rely on the generosity of all the grant giving bodies, organisations and individuals who have funded our work: I would like to say thank you to them all.

Maggie Ponder

Introduction

During the year genetic issues occupied the media, the political scene and the attention of many in the academic, clinical, commercial and regulatory communities.

There was intensive debate about the possible impact of new knowledge for medicine and for society as a whole on a wide range of issues, including genetics and insurance, the use of embryonic stem cells for research and the development of treatments for serious and incurable diseases and the transfer from the research lab into clinical practice of new discoveries – often arising out of projects funded by some of the member charities of the Genetic Interest Group.

In all these and in many other issues too, GIG was an active participant, putting forward the views and opinions of its members and actively campaigning to ensure that the viewpoint of individuals and families affected or at risk from genetic disorders is recognised and responded to.

Throughout the year we have been grateful for the active support of many individuals and organisations who have contributed actively to our work, helping to establish a perspective on the issues that is patient centred, robust and which carries the support of those from whose viewpoint we speak.

The Genetic Knowledge Parks

Early in the year the Secretary of State for Health made a speech in Newcastle in which he announced funding for a network of Genetic Knowledge Parks. These were to be collaborations between universities, the NHS, the commercial sector and patient and consumer interests. They are intended to ensure there is public understanding of the benefits of genetics for health and well being and that the results of British research are used in ways that benefit British patients, the NHS and the economy to the fullest possible extent.

Many of the consortia which put together proposals approached GIG to act as a partner in their application. They recognised that GIG is the only patient organisation that is able to cross the spectrum of genetic disease – from the very common to the very rare and represent issues from a patient and family perspective in a way that carries authority.

Of the successful applicants, five out of six consortia included GIG as a partner – the only patient organisation to be formally involved in this way.

The consortia of which GIG is a formal partner are in London, Cambridge, Oxford, Wales and the NorthWest. We will also be developing links with Newcastle (the sixth Knowledge Park). As a result there will be new posts created in the NorthWest, in Wales

and in London to support public and patient involvement in the new genetics. These will be starting in the autumn of 2002 and will quickly make a difference to public appreciation of the benefit of genetics in medicine.

In addition, there will be a new post in Birmingham. This is funded by a parallel initiative of the Department of Health and again we were delighted to be a partner in this work.

Genetics and the NHS

Surveys undertaken by GIG have shown that there is considerable regional differences over which tests get taken up by the NHS as part of its clinical service delivery and how quickly the new results can be used to benefit patients. This has resulted in considerable variations across the country – with access to help being based in some cases on where you live rather than your clinical needs.

Working closely with colleagues from clinical genetics and alongside laboratory scientists and representatives of member groups, GIG has sought to persuade the Department of Health to address this inequity. We were delighted to be asked to join the Genetics Commissioning Advisory Group (inevitably shortened to GenCAG) when it was set up to address this issue. The early work of this group has been focused on establishing what is done, where and for who and exploring notions of how the system might be made to

“Achieving Partnership”

work more rationally and efficiently in the future. Now this groundwork is out of the way the possible shape of a nation-wide genetic testing network is beginning to emerge and it should be possible to make significant progress towards achieving this in the coming year.

Elsewhere we have worked closely with colleagues to improve understanding of the needs of patients and families for early, accurate diagnosis and the provision of services based on up to date scientific information and best clinical practice. We have organised seminars and workshops and spoken at a wide range of meetings and conferences, always seeking to represent the needs of our members and the families they support in order to achieve change that is useful and useable for the benefit of those in need.

Genetics and Ethnic Minorities

Pritti Mehta, our action researcher has made a significant contribution to the development of understanding about why those from ethnic minority groups often find access to the NHS more difficult than it needs to be – and why those working in genetics centres can sometimes find it difficult to reach out to all sectors of the community, often despite their best efforts to do so. Issues such as the right way of approaching what are thought to be sensitive issues – such as asking about ethnicity, how and where to record this information and what inferences can legitimately be drawn from it (rather than assumptions made which might simply be a result of

prejudice or a lack of knowledge) are all important in ensuring that the NHS is able to meet the needs of all citizens in our multi-ethnic society.

Following a very successful workshop in Leicester attended by representatives of almost all the UK’s clinical genetics centres, Pritti is now working hard to set up some pilot studies in Birmingham and London to look at different ways of asking for and recording information and how it can subsequently be used to improve the quality of genetic counselling for families from ethnic minority groups.

NHS Lifehouse

It is a common experience to go to the doctor and find that the information needed to make proper use of the consultation is lost or hasn’t turned up or that the file is with someone else. This is a particular problem when the condition affecting you is rare (so it is not well understood) or it involves a large number of different clinical specialisms. Computers are an obvious way of solving this problem, but the NHS has a somewhat chequered history of introducing information technology for patient benefit.

Nevertheless, without computers and the creation of an "Electronic Health Record" or EHR, the information held about patients in different places in the NHS will never be linked up. As a result

patients will continue to get a service that is disjointed, not as good as it could be and (especially for those with rare, complex genetic disorders) sometimes plain inadequate.

NHS Lifehouse is a major project designed to address just this problem. On the one hand it is creating a system which will link up patients records wherever they are held in the NHS in a user friendly, accessible way, so that patients and doctors can be confident that the information that decisions are based on is as accurate and as complete as possible. On the other hand developments in the National Electronic Library for Health will make sure that information about the disease is also accurate, comprehensive and complete. GIG is taking a leading role in consulting the public, patient groups and professionals about how they would like to see this organised and just as important, how they want to prevent sensitive personal information from being seen and used by those who should not have access to it – whether they are inside the NHS or external to it.

Thanks to input from GIG (amongst others) this project is focused on patient benefit – either through better services for individuals and families now or through improved support opportunities for doing research or for planning services in the future. Unlike many other attempts to change the way in which the NHS manages information, computers are the means, not the end in this – with the key questions being "What do we need? How can we do it?" rather than "What does this do? How can we use it?"

GIG and Research

Although there has been much progress, sadly it is still the case that most genetic disorders remain incurable and that the only way this will change is through research. Throughout the year we have been working with some of our member groups to lobby the Department of Health, the Medical Research Council and others to commit themselves to ear-marked funding for investigating issues relating to rare disorders.

It is not only the lack of money that is a problem when undertaking research in rare genetic disorders. Too often the system seems designed to create problems rather than encourage much needed investigations to be done. GIG convened a very successful workshop to explore these issues at the MRC. It was attended by researchers, funders, ethicists and patient group representatives. Discussion of the issues raised is ongoing, with hopefully prospects of change that will provide the necessary degree of protection for individuals and families, whilst at the same time encouraging and supporting researchers.

The most controversial area of research development that GIG has been involved with has been the use of embryonic stem cells. Despite the overwhelming vote in favour of allowing this in both the Commons and the Lords, the issue is still causing controversy.

“Achieving Partnership”

GIG organised a European Workshop on this subject in partnership with the European Federation of Biotechnology Task Group on the Public Perception of Biotechnology. This multi disciplinary workshop produced a report that was supportive of this work. At the same time it acknowledged the sensitivities that some may have about this work and suggested practical steps that could be taken to recognise these. The report went to the European Parliament and was used as a source document for a major conference on this issue organised by the European Commission. GIG provided one of the keynote speeches for this event, which was attended by over 600 delegates, representing all shades of opinion from across the whole of the European Union.

GIG has also maintained close links with Biobank UK – the giant study funded by the Wellcome Trust and the Medical Research Council - which will identify links between specific single nucleotide polymorphisms and common disorders such as cancer, heart disease and diabetes.

GIG and the Media

Throughout the year we have continued to act as a source of information and comment both behind the scenes for journalists preparing articles and stories and on the record in the press and on television and radio. We have been extensively quoted in the broadsheet press and in a wide range of popular and specialist journals. In addition we have published a number of longer articles

in more academic journals such as "Community Genetics" and "EMBO Reviews" when we have had the opportunity to explore particular issues in more depth.

The Royal Institution (one of the oldest learned societies in the UK) has established a "Science and the Media Centre" which aims to provide a rapid, accurate response to scientific issues that come up in the press or on TV and Radio. GIG is an active member of this centre and has been called on to provide comment (usually at very short notice) on a number of issues relating to genetics and health. As this becomes established, so the pressure to respond is likely to increase and we look forward to rising to this challenge in the future.

GIG and Europe

In addition to the input to the stem cells issue mentioned above GIG has been active in Europe through its membership of the European Alliance of Patient and Parent Support Groups for Genetic Services in Medicine (EAGS). The principal activity has been through our membership of the Orphan Medicinal Products Committee of the European Agency for the Evaluation of Medical Products, (based in Docklands). This committee is responsible for recommending the granting of "Orphan Designation" for products which are intended to be for the treatment of rare disorders (most of which are genetic). In its first year it was anticipated that about 10 designations would be granted, but the actual number was nearer

70 and the rate of progress has continued to increase. The regulations have proved to be an important stimulus for companies to get interested in developing these products. Already a number of them have been put on the market, with patients starting to benefit.

In addition a successful conference on "Getting Involved in Research" was organised and a report produced which has gone to over 500 patient groups and their umbrella bodies, as well as to researchers, the European Commission, the Department of Health in the UK and many other official bodies in both the public and private sectors.

GIG and its Members

It is a privilege to be able to work for an organisation that draws its strength and its legitimacy from its ability to speak from the standpoint of individuals and families affected by genetic disorders. To do this successfully we need to be confident that the issues we take up and the viewpoints we articulate will have the support of our members and the families they represent.

This is not easy in the field of genetics and health, where new knowledge challenges long held assumptions very frequently and where the issues raised may be technically or ethically complex. All GIG members are very busy with their own issues and finding time and resources to address the wider policy questions can sometimes

be difficult. We are very conscious of the need to make it as easy as possible for our members to consider the issues and give us their views. Thanks to funding from the Baring Foundation we have been able to consult with many of our members about making it easier for members to get involved with GIG and the issues it takes up. One of the most important developments resulting from this consultation has been the decision to appoint a new member of staff to work more closely with members and to improve our consultation and communication. Hopefully the benefits of this investment will be quickly felt once this new post is filled.

GIG and Money

The year started with GIG facing the need to secure new funding to replace existing grants that were coming to an end. Like most small charities GIG is dependent on being able to secure regular injections of money from new funders, as it does not have substantial reserves that it can fall back on if income takes a dip. Fundraising for GIG proved exceptionally difficult in the early part of the year, with many of those we approached endorsing the quality of the proposals we submitted but having insufficient resources to fund them. This was stressful for all concerned. Fortunately the situation improved later in the year and new funding opened up enabling us to finalise the year in a much more confident frame of mind than we started.

“Achieving Partnership”

	Total funds 2002 £	Total funds 2001 £
Incoming Resources		
Membership	9,097	8,621
Grants and Donations	199,261	166,264
Meeting receipts	84	5,023
Sundry Income	107	52
Bank Interest	4,873	7,257
Total Incoming Resources	213,422	187,217
Resources Expended		
Direct charitable expenditure	136,431	150,747
Fundraising	68	12,398
Management and administration	36,227	32,814
Total Resources Expended	172,726	195,959
Net Income (Expenditure) before transfers	40,696	(8,742)

These figures are extracted from the audited accounts for the 2001/02 financial year. A copy of these are available from the GIG

office on request (A4 SAE please).

We are grateful to all our donors, to our member groups and to everyone who has supported us financially or with help, encouragement and advice that gives us the means, confidence and the opportunity to carry on.

Our major donors during the year are: -

ABI
 ABPI
 Ares Serono
 The Baring Foundation
 The D J Fielding Trust
 DEBRA
 Department of Health S64
 European Commission
 EFAM
 Englehorn Foundation
 Glaxo Smithkline Plc
 HSA Charitable Trust
 Lloyds Charitable Trust
 Medical Research Council
 National Lottery Charitable Board
 NHS Lifehouse Project
 Novartis
 Pfizer

PPP Medical Health Trust
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Maggie Ponder (Chair)
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Fiona Helmsley
Ann Hunt
David Watters

Observers

Shirley Hodgson

Director

Alastair Kent

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John Gillott (Policy Officer)
Pritti Mehta (Action Researcher)
Saskia Ottignon (Administrator)

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Professor Timothy M Cox, MD, FRCP, FmedSci, FRCP(Glas), FRSE
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Professor M Patton, MA, MB, MSC, FRCP, FRCPCH

Professor Marcus Pembrey, MD, FmedSci, FRCP, FRCPCH, FRCOG
Professor Sir D Weatherall, FRS, FRCP, FRCPath

Members

Aarskog Syndrome Support Group (UK)

Adrenal Hyperplasia Network

Albinism Fellowship

Alpha 1 Support UK

Alstrom Syndrome UK

Alzheimer's Society

Androgen Insensitivity Syndrome Support Group

Anorchidism Support Group (ASG)

Assert

Association for Glycogen Storage Disease (UK)

Ataxia UK

Ataxia - Telangiectasia Society

ATRX Support Group

Adrenoleukodystrophy Family Support Trust

ARC

Bannayan-Zonana Support Group

Batten Disease Family Association

Beckwith-Wiedmann Support Group

BEHCETS Organisation

The British Porphyria Association

British Heart Foundation

Candid

Cancer Research UK

Cardiomyopathy Association

CGD Research Trust

Children's Mitochondrial Disease Network

CLAPA Reading

CLIMB

Cohen Syndrome Support Group

Congenital Adrenal Hyperplasia Support Group

Costello Support Group

Craniofacial Support Group

The Cogent Trust

CDLS Foundation

Cri Du Chat Syndrome Group

The Cystic Fibrosis Trust

Dariers Disease Support Group

DEBRA

Diabetes UK

Down's Heart Group

Dyskeratosis Congenita Society

Ectodermal Dysplasia Society
Ehlers-Danlos Support Group
Euro-Ataxia
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
Klinefelter's Syndrome Club UK

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
The Neurofibromatosis Association
Niemann-Pick Disease Group
NSPKU
Nystagmus Network

OSCAR

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Osteopetrosis Support Trust

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

Restricted Growth Association

British Retinitis Pigmentosa Society

The Retinoblastoma Society

Rett Syndrome Association UK

Scottish Huntington's Association

Seriously Ill for Medical Research (SIMR)

East London Branch Sickle Cell Society

National Sickle Cell Programme

SOFT UK

SPECS

Sussex ADHD Support Group

STEPS - The National Association for Children with Lower Limb

Abnormalities

Stickler Syndrome Support Group

Shwachman Diamond Support

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 Support Group



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