



# Annual Report 2002 - 3

**Half a Century of Progress**

## Chair's Foreword

This has been a good year for GIG. We have seen the expansion of the GIG team allowing us to reach out into the regions much more effectively. We now have posts in Wales, Birmingham, and Manchester as well as extra posts in the London based team and I would like to formally welcome all the new members of staff. The benefits of the expanded team should start to take effect in the coming months. In the future we aim to have more regionally based staff with Scotland being the first priority.

The work done by GIG continues to be very diverse but always with the central theme of promoting and highlighting issues of concern for people living with genetic conditions. As genetic technology is seen as becoming more relevant to more people it is vital that we continue to focus on concerns relevant to our members whilst at the same time keeping abreast of issues of interest to the wider public. Getting this balance right is difficult; however, our Director Alastair Kent has done so successfully this year and I have confidence that he will continue to do so in the future.

The team works extremely hard on our behalf and I would like to thank each of them for their contribution. I would also like

to thank all the trustees for their support during this year. Without money GIG could not work. This year we have been more successful than we were in the previous year with raising funds and I would like to take this opportunity to thank all the grant giving organisations and individuals who have contributed so generously.

Maggie Ponder  
September 2003

## High Points of 2002-2003

Even in a very busy year, some things stand out as high spots – achievements we are particularly proud of, or events that have pleased us. We have picked some of these out, to give a quick overview of 2002-3.

During the year we:

- ✓ Improved and increased contacts between GIG and our members
  - ✓ Re-designed our web-site
  - ✓ Re-vamped our quarterly newsletter, GIG Today
  - ✓ Produced attractive and informative publications
  - ✓ Organised a variety of inputs from our members to a number of research projects.
  - ✓ Attracted over 50 GIG members to take part in the Royal Society's "People's Science Summit"
  - ✓ Steadily increased our membership
  - ✓ Improved our fund-raising
  - ✓ Carried out successful pilot schemes for ethnicity monitoring in NHS Clinical Genetics Centres.
- ✓ Helped to persuade Government to invest in gene therapy
  - ✓ Recruited new workers in Wales, the North West and Birmingham
  - ✓ Appointed an Assistant Director
  - ✓ Organised consultations and created responses to many Government initiatives and policies
  - ✓ Appeared and commented regularly in the press and on radio and TV
  - ✓ Increased our input to the Human Genetics Commission
  - ✓ Took part in campaigns to influence the British and the European Parliaments about issues important to GIG's members
  - ✓ Contributed to a range of national and international conferences, workshops and seminars

*and much more!*

## Introduction

When James Watson and Francis Crick wrote their famous letter to Nature announcing the structure of DNA they could not have realised just how much impact their discovery would have. The knowledge that has flowed since that letter was published and the techniques that have been invented to unravel the secrets of the double helix have enabled scientists to understand what goes wrong at the molecular level when mutations occur that result in serious genetic diseases.

This is an incredibly powerful tool, and in the last few years we have begun to see significant practical benefits for individuals and families affected by a growing number of different genetic diseases. The pace of change seems to be picking up, and we hope that more and more conditions will become amenable to treatment as scientific research reveals the secrets locked up in the genetic code.

We were greatly honoured by the fact that James Watson agreed to contribute a personal foreword to one of GIG's publications produced to commemorate the 50th anniversary of the announcement of the double helix. This was very well received, and has been widely distributed and downloaded many times from our web-site ( [www.gig.org.uk](http://www.gig.org.uk)).

Elsewhere, as this report shows, GIG has been active on behalf of its member groups and the families that they support in many spheres, pushing hard to ensure that the interests of those who live with the daily impact of serious genetic disorders are heard by those with the power and resources to achieve change.

As always, GIG cannot operate in isolation. We are dependent on and very grateful for the support and the encouragement we receive from our members, from the scientists and the doctors we work with, and from those who fund and support us in many other ways.

## The Genetic Knowledge Parks

During the year the Knowledge Parks have made real progress, and are already having an impact on the opportunities to develop and improve services. Appointments to the posts that have been created have been made, and we can see substantial benefits on the horizon as a result of this initiative.

In London, Pritti Mehta is working with colleagues in the IDEAS Genetic Knowledge Park to produce literature that will open up genetic services to families from minority ethnic

communities. GIG also played a significant role in a major conference at the Science Museum organised to launch IDEAS that was attended by over 150 people from all walks of life.

Buddug Williams, the GIG Cymru worker in Cardiff has begun to develop a network that will help the development of the All-Wales Clinical Genetics Services. The Welsh Assembly Government has voted a substantial (£1.5 million p. a.) increase in the budget for clinical genetics, and GIG is working alongside the clinicians to make sure this is used to good advantage in ways that meet the real needs of patients and their families.

NoWGen, the North West Genetic Knowledge Park, will have a public access area and an information centre in its new building when it opens. Providing the public face to this will be Jo Ayres, the Information Officer. She will be backed up by Stuart Nicholls, our Research Fellow, who will be responsible for ensuring that the research programmes have a proper patient and user input to their planning and delivery.

Getting research into rare genetic disorders underway is difficult at the best of times. A proper regulatory framework gives confidence to researchers and provides protection for patients and families. GIG has been working with the Oxford Genetic Knowledge Park to try and create such a framework

something we are hoping will be taken up by research ethics committees in the future. The Knowledge Park in Cambridge has professional education as one of its foci. Volunteers from some of our member groups have played a significant role in shaping recommendations for the education of a wide range of different medical professionals and managers. This project is led by Hilary Burton, a public health doctor in the Cambridge GKP who is a cooptee to the GIG trustee board.

Although Birmingham does not have a Knowledge Park as such, the Genetic Education Unit in the Genetics Centre has been very active, and Anna Lane, the GIG appointee, has been busy developing links with schools – including organising an event for the Human Genetics Commission's members to meet a cross section of the City's school children when they visited the city for one of their regular meetings.

To oversee these developments and to strengthen GIG's management team we have appointed a new Assistant Director, Tom Barclay.

## Genetics and the NHS

For much of the year the genetics community held its breath as the Department of Health put together a green paper that would lay out its vision for the future of genetic medicine in the NHS. Initially expected in the autumn of 2002, it actually arrived in the summer of 2003. During its gestation it had become a white paper – a much more powerful statement of policy and direction from the Government. Approximately £50 million of new initiatives were contained in the white paper, laying some good foundations on which to build a modern, equitable and responsive service for the individuals and families who need the care and support the NHS provides.

Elsewhere the Genetics Commissioning Advisory Group and the UK Genetic Testing Network Steering Committee – official Department of Health bodies on which GIG's an active member – continue to grapple with the complexities of creating a rational and fair structure for patient access to services across the NHS, irrespective of geography and other non-clinically relevant factors. Looking at the NHS from our members' perspective, it feels like trying to turn a supertanker, in that it seems like ages before the movement of the steering wheel on the bridge results in any perceptible

change in direction. However, GIG's input to the process has been significant – particularly in the area of communicating the results of tests to patients and families, and the incorporation of guidance on good practice in the official documentation put out to the Commissioners of Services, and the NHS Trusts which provide them.

We continue to be active in pressing for more and better services for families at risk, and the elimination of the inequalities and variations that still exist today.

## GIG and Research

In addition to our work in partnership with the Oxford Genetic Knowledge Park GIG has played an active role lobbying for more funding for research. One of our founder trustees, Ann Hunt, has been a member of the Gene Therapy Advisory Committee for the last six years. She observed that most of the applications for approval of research projects coming forward were for the treatment of common complex disorders like cancer. The rare single gene conditions did not seem to be getting a look in. A survey by Ann confirmed that there was a real problem with funding, and we worked to persuade the Department of Health to recognise this. Ann's input was crucial to the success of this, and we were delighted

to see the announcement of a £3 million fund earmarked for this in the White Paper, and of a further £2.5 million for gene therapy research into cystic fibrosis as well.

The UK Biobank project started in earnest during the year with the appointment of the first chief executive (Dr. John Newton). GIG organised a very successful consultation with GPs in North London designed to set out the issues that they would want to see incorporated into this £60 million project for them to be interested in helping to recruit their patients to participate. We continue to work closely with UK Biobank to develop its public consultation strategy, and to promote understanding of what this venture will and will not do. In the long term, the benefits to families that will result from greater appreciation of genetics in primary care will be very important for all GIG members.

GIG is also working with the School of Health and Related Research at Sheffield University to investigate ways of supporting GPs so they can help families with rare genetic disorders more effectively. We are recruiting volunteers willing to share their experience and expertise so that recommendations for change are firmly grounded in real experiences.

## **Equity, Ethnicity and Access to Services**

As well as working with colleagues from the London Ideas GKP, Pritti Mehta has been busy completing a project for the Department of Health on the development of a monitoring framework for recording ethnicity data for clinical genetics services. This information is important for two main reasons – because without it the Health Service will not know if all citizens are accessing its services, and because ethnicity data enables geneticists to improve their clinical practice – giving quicker, more accurate diagnoses in ways that are sensitive to the cultural traditions and beliefs of the different communities in multi-ethnic Britain. The report of this project is available on the GIG web-site. We are in discussion with the Department of Health as to the next steps – how to take the lessons learned out into all regional genetics centres, and also to the wider NHS.

## **Communicating with the Public and the Media**

Genetics continue to be a “hot topic”. Stories of developments (and what are often made out to be “disasters” too) crop up almost every week, and GIG has been constantly on the alert for opportunities to remind readers and viewers of the needs and opinions of people who live with the impact

of genetic disorders on themselves and their families every day. Comments and opinions from GIG staff and trustees, and from individuals with direct personal experience of issues under discussion have appeared regularly in the press, on TV and radio and in specialist journals and magazines here and around the world. In addition GIG staff have published a number of articles in professional journals and other specialist publications.

GIG's web-site developed and improved during the year thanks to a grant from the Department of Trade and Industry and other funders it has proved an invaluable tool in getting our views across. Visited by over 10,000 people every month it provides a resource for individuals, families, policy-makers, teachers, journalists and many others. Visitors to the site can get information on a wide range of topics, download GIG's publications, read our responses to government and other consultations and link with many other relevant web-sites, providing a gateway to today's issues in genetics from the perspective of patients and their families.

## **GIG and its Members**

Thanks to the support of the Baring Foundation last year GIG surveyed its member organisations to find ways in which we could provide a better opportunity for them to contribute to the issues which shape the services and support they receive (or would hope to!).

As a result of the feedback we received we have appointed Melissa Winter as our Membership and Communications Officer. She has made a dramatic impact already, taking day to day responsibility for our web-site, and completely re-vamping and improving our newsletter, "GIG Today", and our other publications. In addition she is in regular contact with members on a wide range of issues – seeking opinions, recruiting volunteers for media interviews or to participate in research projects, and generally keeping people informed about what is going on.

Without the endorsement and support of our members, GIG would not be able to achieve anything. It is both a privilege and a responsibility to ensure that we do the best we can to make their views and needs known to all who need to hear them.

## European Issues

Once again the dominant issue has been Orphan Medicinal Products. The committee that recommends orphan designation has been up and running for three years now, and GIG's Director is one of the patient representatives with a seat at the committee table. At the end of April 2003, the number of applications which had been considered for orphan designation was 244, of which 137 had been recommended for designation, and 8 of which had gone on to be licensed as drugs for treating rare diseases. The numbers are rising, but getting a drug onto the market is only the half way point in its journey to the patient. The NHS must be willing to provide and pay for these new treatments, and doctors must be aware of them so they can be prescribed. We have begun working with commissioners to address this issue in a fair and logical way so that those who need to benefit can be confident that they will do so – and as soon as possible after a new treatment is licensed.

## Genetics and Insurance

Any discussion of genetics and insurance runs the risk of generating more heat than light, as it is an issue that raises strong passions on all sides. Following our usual policy of

trying to bring different factions together, and to look for areas of agreement as well as seeking to understand the reasons why disagreement occurs, GIG has convened a series of workshops that provide a forum for the discussion of potentially contentious issues in a way that promotes understanding rather than the repetition of pre-existing positions. The reports of these meetings – on “Insurance and Rare Events,” “Family History”, and “The Nature of Risk,” are on our web-site. They have been more than mere talking shops. One practical outcome has been agreement by the Association of British Insurers to develop “templates” for the systematic collection of accurate information about rare genetic diseases, so improving the quality, logicity and fairness of decisions they make as a result. The process is under way – we will continue to monitor the situation and make inputs where they are needed to keep it on track.

GIG is also a member of the UK Forum on Genetics and Insurance; John Gillott, our Policy Officer, spoke at their Annual Conference.

## Setting the Policy Agenda

Increasing attention is being paid to input from patients and the wider public in the NHS, and across the range of

Government initiatives where genetic developments have an impact on health care and public policy. GIG has a place in many of the most important meetings where policy decisions are made - in the committees of the Department of Health and of the Medical Royal Colleges, on the Advisory Boards of the Association of British Insurers, and the Boards of many of the genetic Knowledge Parks, to name but a few. The Director was also appointed a member of the Human Genetics Commission (Philip Webb, one of GIG's trustees, has been a member since the start). Ann Hunt's membership of GTAC has been mentioned on page 6, whilst other committees and boards have benefited from the impact of GIG staff and trustees, and from that of our member groups too.

In addition to this formal involvement, GIG has been active informally, briefing MPs, officials and journalists on issues ranging from embryonic stem cells to over the counter testing, sex selection and the future supply of scientists for genetic testing laboratories. Wherever there is an opportunity to ensure that things which will impact on our members and their families for good or ill are understood and appreciated, so that desirable outcomes can be maximised, and undesirable ones avoided as far as possible, GIG will continue to offer its input.

## GIG's Money

In 2001-2 GIG's finances were on a roller-coaster. Although we finished the year in a relatively healthy position we went through a difficult period during the year. This year things have been more stable, thanks to funding from the Knowledge Parks and the continuing support of our regular funders in the public and the private sectors and the generosity of new donors. However, there is a constant need to secure new income, and to guard against complacency. We are careful to try and ensure that every penny is spent to good effect, and because of this are confident that we can give a good account of ourselves to anyone who chooses to ask. Nevertheless, it remains the case that with more, and more secure funding, we could do more, and as a result improve the situation for our members and their families.

As always, GIG is careful to preserve its independence and its autonomy. Money which might be offered with unacceptable conditions attached would be refused. GIG sets its agenda and its strategy, then seeks the resources to put this into practice. We will not allow ourselves to be led by money into positions which are not in the interests of those living with genetic disorders, their families and the groups they choose to join.

**Extract of Audited Accounts**

Incoming Resources	Total Funds 2003 (£)	Total Funds 2002 (£)
Membership	17,350	9,097
Grants and Donations	259,957	211,035
Meeting receipts	-	84
Sundry income	439	107
Bank interest	6,459	4,873
<b>Total Incoming Resources</b>	<b>284,205</b>	<b>225,196</b>
Resources Expended	(£)	(£)
Direct charitable expenditure	191,243	136,431
Fundraising	5,010	68
Management and Administration	37,929	36,227
<b>Total Resources Expended</b>	<b>234,182</b>	<b>172,726</b>

**Major Donors**

	£
ABPI	7,500
Amersham Plc	1,500
Bio-Industry Association	5,653
Birmingham Women's Hospital	4,000
Department of Health (core grant)	25,000
Department of Health (project grant)	42,000
DJ Fielding Charitable Trust	10,825
European Union	27,351
London IDEAS Genetic Knowledge Park	11,320
Medical Research Council	15,000
Motor Neurone Disease Association	2,000
NoWGen Genetic Knowledge Park	10,000
Oxford Genetic Knowledge Park	5,000
Pfizer Ltd	10,000
Roche Products Ltd	6,999
The Mercers Company	1,000
Wellcome Trust	19,429
West London database project	55,000
Other Donations	380
<b>Total</b>	<b>259,957</b>

**Members:**

Aarskog Syndrome Support Group (UK)  
 ADHD National Alliance  
 Adrenal Hyperplasia Network  
 Adrenoleukodystrophy Family Support Group  
 Albinism Fellowship  
 Alkaptonuria Society  
 Alpha 1 Support UK  
 Alstrom Syndrome UK  
 Alzheimer's Society  
 Androgen Insensitivity Syndrome Support Group  
 Aniridia Network UK  
 Anorchidism Support Group (ASG)  
 Anti Natal Results and Choices  
 Angelman Syndrome Support and Education Research Trust  
 (ASSERT)  
 Association for Glycogen Storage Disease (UK)  
 Association of Multiple Endocrine Neoplasia Disorders  
 (AMEND)  
 Ataxia - Telangiectasia Society  
 Ataxia UK  
 ATRX Support Group  
 Batten Disease Family Association  
 Beckwith-Wiedemann Support Group  
 BEHCETS Organisation

British Heart Foundation  
 British Porphyria Association, The  
 British Retinitis Pigmentosa Society  
 Cancer Research UK  
 Cardiomyopathy Association  
 CDLS Foundation (Cornelia de Lange Syndrome Foundation)  
 CGD Research Trust (Chronic Granulomatous Disorder)  
 Children's Mitochondrial Disease Network, The  
 Children Living with Inherited Metabolic Disorders (CLIMB)  
 CMT UK  
 Cogent Trust, The  
 Cohen Syndrome Support Group  
 Congenital Adrenal Hyperplasia Support Group  
 Contact a Family (Scotland)  
 Costello Support Group  
 Cri Du Chat Syndrome Group  
 Cystic Fibrosis Trust, The  
 Darriers Disease Support Group  
 DEBRA (Epidermolysis Bullosa (EB)  
 Diabetes UK  
 Down's Heart Group  
 Dyskeratosis Congenita Society  
 Dystonia Society, The  
 East London Branch Sickle Cell Society  
 Ectodermal Dysplasia Society  
 Ehlers-Danlos Support Group

Eyeless Trust, The  
 Fragile X Society, The  
 FSP Support Group  
 Fuchsfriends UK  
 Galactosaemia Support Group  
 Gauchers Association  
 Gene Aid  
 Gorlin Syndrome Group  
 Gwent Huntington's Disease Association Support  
 Haemochromatosis Society  
 Haemophilia Society  
 Headlines Craniofacial Support  
 Heart Haven  
 HITS (UK) Family Support Network  
 Huntington's Disease Association  
 Huntington's Disease Association (Colchester Branch)  
 International Glaucoma Association  
 International Autistic Research Organisation, The  
 Jennifer Trust for Spinal Muscular Atrophy, The  
 Klinefelter Organisation (UK)  
 Laurence Moon Bardet Biedl Society  
 Long QT Family Support Group  
 Macmillan Cancer Relief  
 Manchester Sickle Cell and Thalassaemia Centre  
 Marfan Association UK  
 Motor Neurone Disease Association

Muscular Dystrophy Campaign  
 Muscular Dystrophy Support Group  
 Myotonic Dystrophy Support Group  
 Narcolepsy Association – UKAN  
 NASPCS  
 National Federation for Families  
 with Visually Impaired Children (LOOK)  
 National Sickle Cell Programme  
 Neurofibromatosis Association, The  
 Niemann-Pick Disease Group  
 National Society for Phenylketonuria UK (NSPKU)  
 Nystagmus Network  
 OSCAR  
 Osteopetrosis Support Group  
 Pemphigus Vulgaris Network  
 Peutz Jeghers Syndrome Support Group  
 Polycystic Kidney Disease Charity (PKD)  
 Prader Willi Syndrome Association UK  
 Primary Ciliary Dyskinesia Family Support Group  
 Primary Immunodeficiency Association  
 Progressive Supranuclear Palsy [PSP Europe] Association  
 Psoriatic Arthropathy Alliance  
 PXE International  
 The Pseudoxanthoma Elasticum (PXE) Support Group (PiXiE)  
 Restricted Growth Association  
 Retinoblastoma Society, The

Rett Syndrome Association UK  
 Sudden Adult Death Trust (SADS) UK  
 Scottish Huntington's Association  
 Seriously Ill for Medical Research  
 Shwachman Diamond Support  
 Sickle Cell and Thalassaemia Service  
 Society for Mucopolysaccharide Diseases  
 SOFT UK Support Organisation For Trisomy 13/18  
 and related disorders  
 Specific Eye Conditions (SPECS)  
 STEPS - Association for People with Lower Limb Abnormalities  
 Stickler Syndrome Support Group  
 Syndromes without a name (SWAN)  
 Tay Sachs and Allied Diseases Association  
 Telangiectasia Self Help Group  
 Tuberous Sclerosis Association  
 Turner Syndrome Support Society (UK)  
 UK Thalassaemia Society  
 Unique- The Rare Chromosome Disorder Support Group  
 Williams Syndrome Foundation Limited  
 Worster Drought Syndrome Support Group  
 XLH Network (X-Linked Hypophosphatemia)  
 XP Support Group

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John Gillott - Policy Officer  
Pritti Mehta - Project Manager  
Lucy Ullmann - Administrator  
Melissa Winter - Membership and Communications Officer  
Anna Lane - Development Officer (based in Birmingham)

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**Unit 4D, Leroy House  
436 Essex Road  
London NI 3QP**

**Tel: (020) 7704 3141**

**Fax: (020) 7359 1447**

**Email: [mail@gig.org.uk](mailto:mail@gig.org.uk)**

**Website: [www.gig.org.uk](http://www.gig.org.uk)**

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