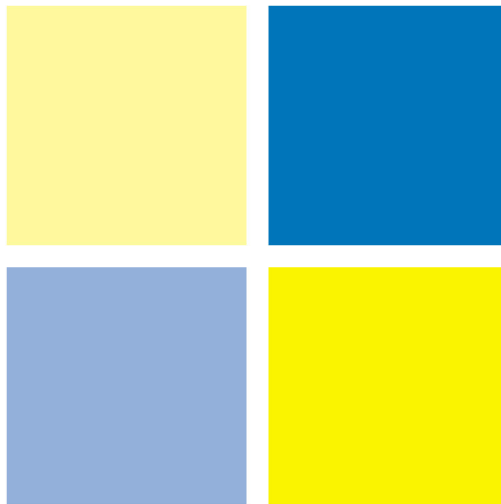


Unique – Rare Chromosome Disorder Support Group
Annual Report
1 April 2004 to 31 March 2005

Unique

**ANNUAL REPORT
31 MARCH 2005**



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Letter from Group Coordinator

Unique has had yet another very successful year which I have to attribute to the dedication and hard work of its employees and to the continuing support of our members, their families and friends.

It is quite a staggering statistic that a rare chromosome disorder appears in at least one of every 200 births, which is why **Unique** continues to evolve and seek to meet the needs of every family that is affected not only here in the UK but in 64 countries worldwide.

I hope in reading this report you can see how the funds we have managed to secure over the year have been put to good use and with further funding we can continue in our work and help others who find themselves thrown into a world where their child really is **Unique**.

Edna Knight

ANNUAL REPORT

The charity was established on 24 July 1993.

Change of Legal Status

During 2004, the committee undertook lengthy discussion over converting to an incorporated body. At the time of this report, the Management Committee have received a Notice of Formal Resolution to change the legal status of The Rare Chromosome Disorder Support Group (RCDSG) to become an incorporated company and retaining its charitable status by becoming **Unique** RCD Ltd. The date of agreement is set for 23rd April 2005.

OBJECTIVES

The Rare Chromosome Disorder Support Group, normally known by its short name of **Unique**, aims to provide links between families whose children have similar clinical and/or practical problems caused by rare chromosome disorders. A rare chromosome disorder causing disabilities and health problems can occur in at least one in every 1,000 live births. The overall figure for ANY rare chromosome disorder including balanced rearrangements that do not cause disability and ill-health is at least 1 in 200 live births.

Unique also acts as an international family support group in over 64 countries worldwide.

Unique aims to promote awareness of rare chromosomes disorders both in the UK and abroad and works closely with professional contacts.

ACTIVITIES

Unique has three Trustees and a Management Committee of thirteen members (including trustees), who meet at least four times a year to formulate and review the group's work. To cope with the added workload, **Unique** now employs five members of staff, two full-time and three part-time.

The group's activities are communicated to members by way of:

- individual contact
- local meetings
- a bi-annual conference
- a three times a year newsletter
- a comprehensive website at www.rarechromo.org

A comprehensive database detailing lifetime effects of specific chromosome disorders is maintained, providing an invaluable source of information to new members, existing members and professionals. An increasing amount of assistance to research projects is envisaged, at all times protecting members' confidentiality.

There was no conference during the 2004-5 financial year. Plans are currently in place to organise the next conference in October 2005.

We are all looking forward to 2005 as the charity celebrates its 21st anniversary



The last annual general meeting was held in May 2004 when the previous accounts were submitted and approved by the Committee Members.

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ORGANISATION

As at 31 March 2005, our current resources are as follows:

Trustees

- Mrs Edna Knight
Group Co-ordinator
The group was founded in 1984 by Edna Knight as the 'Trisomy 9 Support group' with just 5 member families. In 1989 the group expanded to include children with any rare chromosomal disorder and in 1993 the group was granted charity status.
- Mrs Gillian Manvell
Gill is an active member of the committee and has been a trustee since 1993. She is also a trustee of other charities and school governor we are very grateful to her dedication to *Unique* and her valuable contribution during the transition period towards *Unique* RCD Ltd.
- Mrs Marie Layng
Marie has been a trustee since 1999. As a language teacher and a Spanish Tutor for the Open University, she has, unfortunately, been unable to attend many of the committee meetings but remains a valuable contributor to all discussion and decisions.

Retired Trustees

- Mr Nigel Barrett
Nigel retired as our voluntary Treasurer and a Trustee after fourteen years service. The committee were sorry to see Nigel leave and recognise the huge impact he made to the day to day running of *Unique*. His contribution and commitment to the group has been greatly appreciated over the years, but we acknowledge that other commitments including his family must come first.

Employed members of staff

- Dr Beverly Searle - Development Director, has now been a full time employee with Unique since April 1999. Her primary role and responsibilities include being our main contact with both new and existing families and the professional medical contacts both in the UK and internationally and also identifying new areas for development to benefit member families. Beverly is also line manager for the Information Officer, Assistant Information Officer and Family Support Officer.
- Mrs Prisca Middlemiss - Information Officer, joined us in June 2003 as a full time employee and is funded by a three year grant from The Health Foundation. Her specific role has been to create family friendly literature which covers specific rare chromosome disorders and also to develop further relationships with professional contacts both in the UK and abroad.
- Mrs Lydia Dickie -Assistant Information Officer, was originally employed under a 12 month part-time contract in 2003. During 2004, we were successful in applying for a grant from BBC Children in Need to cover her salary for a further two years. Lydia's work has been invaluable in extending our knowledge of educational, developmental and behavioural issues and concerns which affect many of our families.
- Mrs Marion Mitchell - Family Support Officer, has now been a part time employee since April 1999. Her primary role and responsibilities cover the main day to day practical issues in running of the group. Marion works closely with the Development Director in supporting families and local contacts and organising the group's conference.
- Mrs Julie Griffin - Treasurer, originally a member of the committee since October 2002, volunteered to be an assistant to the previous Treasurer Mr Nigel Barrett in May 2004. In July 2004, Julie took over the role of Treasurer and we were successful in applying for a grant from the Lloyds TSB Foundation to cover a part time salary to cover the role from October 2004. Julie has a banking and marketing background.

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PROGRESS AND ACHIEVEMENTS

The chart below tracks the continued growth of enquiries we have supported over the past 12 months.

	As at Mch 2004	As at Mch 2005	% Growth in the last 12 months
Members	4,000	4,500	13%
Local contacts	128	141	10%
Telephone enquiries	4,500	5,200	15%
Email enquiries	32,146	49,560	54%
Website 'hits'	87,798	119,569	36%
Messages posted on members forum	2,000	2,352	17%

Events

- Unique family day out**
 Our Family Support Officer arranged for a *Unique* family day out in June 2004 to Whipsnade Wild Animal Park, Dunstable, Beds. It was attended by 20 families from all over the UK. We received several positive comments afterwards, which made it worthwhile.
- Unique Conference**
 The next conference has been booked for 21st-23rd October 2005 at the Daventry Hotel.

Broadening the facilities we offer

- Unique Tales.** A child friendly comic aimed at 7 to 10 year olds to help explain the issue to siblings of affected children. Each

UK member received a copy in June 2004 to share with their own children and also raise awareness with professionals who they may deal with on a day to day basis. Non-UK families and professionals can order copies by emailing us.

- Volume 2** of the **Little Yellow Book** series for adults continued to evolve over the last 12 months and is near final completion. Publication was delayed deliberately in order to be able to include details of new molecular techniques like DNA microarray technology. Illustrations for the book have now been finished and we hope to print in Summer 2005.

Specific to the grant we received from The Health Foundation, our Information Officer has continued to research and work on the publication of specific chromosome disorder leaflets as detailed below:

- Leaflets** on a wide range of rare chromosome disorders have been completed, including:

 - deletions 1p36
 - 1q4 deletions
 - deletion 2q37
 - deletion 3p25
 - 3q2 deletions
 - 4p duplications
 - 5q22 deletions
 - 5p duplications
 - 6p deletions
 - 7q36 deletions
 - deletion 8p23.1
 - 9p deletions
 - 9p duplications
 - 10q 26 deletions
 - 13q deletions
 - Idic 15
 - 17p duplications
 - Jacobsen syndrome
 - Pallister-Killian syndrome
 - Robertsonian Translocations
 - Triploidy
 - Diploidy Triploidy
 - XYY syndrome
 - Tetrasomy X
 - Pentasomy X
 - XXYY syndrome
 - XXXXY syndrome
 - ring 14, 15, 18, 20, 21 and 22
 - Trisomy 8 Mosaicism
 - Trisomy 9 Mosaicism

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- Mosaic Trisomy 16
- Uniparental Disomy 14
- Mowat Wilson syndrome,
They are verified by medical experts, geneticists and Professor Maj Hulten, *Unique's* chief medical advisor.
- **Questionnaires** went out to all *Unique* families affected by all the disorders listed above. The questionnaires supply some of the raw data that feed into the leaflets that are prepared for publication.
- A chance observation that an unusually high proportion of mothers with a son with 49,XXXXY had taken the fertility drug clomiphene led the Information Officer to conduct a **survey** of all fertility problems and assisted reproduction in all *Unique* families with a sex chromosome disorder. The results are still being analysed but the signalling of this type of association is an important use of the *Unique* database. We are not aware of any other chromosome database worldwide that is structured in a way that would allow this type of investigation.
- In November the Information Officer organised a **study day** for families with a daughter with Pentasomy or Tetrasomy X. This was the first disorder-specific day ever organised by *Unique* and the first study day ever run for families affected by these sex chromosome variations. Families came from Ireland, Italy, Switzerland, Holland and Denmark as well as from the UK and for the great majority of people this day offered their first opportunity ever to meet someone else with these extremely rare chromosome conditions. Eighteen families and fourteen affected girls and women came to the study day - five with pentasomy X and nine with tetrasomy X, ranging in age from three to 27 years. As well as offering families the chance to meet and talk and for one brief day to be in a community where tetra and penta X were the norm, four experts in their field led teaching sessions.
- Dr Raoul Rooman, paediatric endocrinologist from Antwerp University Hospital in Belgium, captivated families with his introduction to tetrasomy and pentasomy X (see his presentation on our website). Dr Rooman also launched a

prospective Tetrasomy and Pentasomy X registry at the study with the aim of updating research.

- Clinical geneticist Dr Helen Stewart from Oxford talked about the genetics of extra X chromosomes, Professor David Skuse from the Institute of Child Health in London told families what is known about the behavioural aspects of sex chromosome conditions in girls and women and *Unique* member and Wendy Magee, Parent Partnership Officer for Solihull, explored educational issues.

Improvements of last 12 months

Database update:

- We have re-designed the database so that it can now hold much more information about individual members than it could before. We have also altered the database so that we can now distinguish between members wanting to receive a paper copy of the newsletter and those who are happy to go to "collect" an electronic version of the newsletter from the new newsletter archive held in the password-protected part of the website. So far about 500 member families have chosen the electronic delivery route and this will save the group a considerable sum in postage and printing costs.

Website update:

- As detailed above members can now download a copy of the newsletter.
- A new Fundraising Events page has been set up to advertise events which our members have organised to other members and visitors to the site.
- Under the donations page we have now included a *Unique* Gift aid form which members can download to send with their donations as well as a new regular giving form if members wish to set up a regular standing order for donations.

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AWARENESS RAISING

May 2004

- The Development Director gave a presentation to midwives and a few young cytogeneticists attending the first genetics course run by the London Genetics Knowledge Park at the Institute of Child Health (ICH) in London in May 2004.

August 2004

- The Information Officer attended the biennial meeting of the International Association of Child and Adolescent Psychiatrists. The session of most interest to us was on the developmental and behavioural outcomes of various chromosome disorders, including 22q11 deletions (Ann Swillen, Leuven); Prader Willi (Anik Vogels, Leuven); Angelman (Christine Freitag, Homburg) and 7q11.23 deletions (Gene Fisch, North Shore Research Institute, Long Island). We have corresponded with Dr Fisch about extending his studies on developmental outcomes to some of our families. The other contacts were extremely helpful for the work on the information leaflets.
- Our Assistant Information Officer met with a Clinical Behavioural Therapist, Natasa Momcilvic, to discuss the leaflet on puberty and sexuality. Natasa is also trying to set up parenting classes specifically aimed at parents of children with a learning disability. We also met with Karen Adams, from the Challenging Behaviour Foundation, who is keen to develop contact with *Unique*. The CBF have a very good relationship with the Tizard Centre.

September 2004

- *Unique* had an awareness-raising stand at the British Society of Human Genetics Annual Conference at the University of York in September, manned by Beverly Searle and Prisca Middlemiss, which attracted a lot of attention. The majority of delegates were clinical geneticists, genetic counsellors, genetic nurses, cytogeneticists and molecular geneticists from across the UK. We were also very pleased to have *Unique* member Kathryn McKerracher attend with us on a voluntary basis; Kathryn has a deletion 18q, had recently graduated with a BSc (Hons) in genetics and is now

doing a Masters degree in genetics. Meeting her was a rather salutary experience for many of the geneticists! It was very encouraging how many delegates came up to express their enthusiasm for the work of *Unique*. Information packs about the work of *Unique* were handed out to the majority of the 700+ delegates.

- The Information Officer has represented *Unique* throughout the year on the Cambridge Genetics Knowledge Park's learning disability and genetics project. The work of the public participation initiative of this project, which is now being run temporarily by *Unique* committee member Eleanor Fiske, will lead to the publication of regional recommendations for the management of genetic diagnosis.
- The Assistant Information Officer attended the opening of the new Centre for Autism and Related Disorders at The Institute of Psychiatry, London. Prof. Patrick Bolton mentioned *Unique* in his presentation. She also met with Sarah Curran who has carried out studies on possible links between autism and proximal deletions of chromosome 15q. Sarah plans to continue her research with a study of chromosome 22q13 deletions. Most of the speakers held the view that there was a very strong argument for a genetic link in autism but that it was unlikely to be due to a 'single gene' but possibly to a combination of factors. Sir Michael Rutter firmly believed that it was a multifunctional disorder related to environment and genetics
- As mentioned in our 2004 report we are partners in a pan-European project involving a wide range of participants. This project is called **SAFE** (Special Non-Invasive Advances in Foetal and Neonatal Evaluation) which is a network of excellence involving 50 research groups from across Europe investigating non-invasive methods of testing and diagnosis for chromosome abnormalities both *in utero* and in the neonatal stage. The Development Director and Information Officer attended the first working meeting of SAFE at the University of Warwick in September 2004. They negotiated our exact involvement for the first 18 months in three of the seven work packages. We

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have received funding to cover the costs of this stage of the work.

Knowledge Park at the Institute of Child Health (ICH) in London in January 2005.

October 2004

- Following on from the very positive response of the cytogeneticists at her presentation in May, Dr Beverly Searle was also invited by Dr. Jonathan Waters, Deputy Director of the NE London Cytogenetics Laboratory, to give a presentation about the work of *Unique* to about 50 cytogeneticists and clinical geneticists at their monthly lunchtime lecture slot in October.
- The Development Director attended a day long conference and workshop, organised by the London IDEAS group and the Department of Health, entitled "Genetic Services Evolving to Meet the Needs of Your Local Health Community". She also represented *Unique* at the Genetic Interest Group's annual conference later in the month.
- A long-time and very committed *Unique* member Andy Tickle gave a presentation about the work of *Unique* at the North West Genetic Knowledge Park (NOWGEN) meetings for support groups.

December 2004

- The Development Director appeared on the TV programme BBC South Today and was interviewed on Radio Solent and Three Counties Radio where she spoke about the work of *Unique* and the extraordinary challenges faced by our member families.
- *Unique* was invited by the co-ordinator Professor J-J Cassiman of the University of Leuven in Belgium to become a partner in another successful bid for funding from the EC for a Network of Excellence entitled **Eurogentest** (Genetic Testing in Europe - Integrated Network for test development harmonization, validation and standardization of services). The Development Director attended an initial meeting in Leuven in December 2004. Work on the project began in January 2005, once funding had been received.

January 2005

- Dr Beverly Searle was asked back to give a presentation to the second course for midwives run by the London Genetics

February 2005

- Our Family Support Officer participated in a seminar titled "Parenting and sexual health issues for people with intellectual disabilities" at The Royal Society of Medicine, Wimpole Street, London.

March 2005

- Dr. Beverly Searle was invited to speak to a group of second year medical students from the Royal Free and University College Medical School to describe her personal experiences of living with a rare chromosome disorder in her family

Other

- The Development Director continues to be a patient representative on a clinical group that has been formed to assess individual cases to advise Primary Care Trusts in the South East on the clinical appropriateness of requests for funding for Pre-implantation Genetic Diagnosis (PGD). She has now assessed 13 out of 26 cases put forward for funding. It is envisaged that the experiences of this specialist group can eventually be rolled out nationwide.
- The Development Director has been asked to be on the website development steering group of the rare disorder UK mapping project being organised by the WellChild Trust.
- Following *Unique's* involvement in the Cambridge Genetics Knowledge Park, a representative was invited to sit on the Advisory Board for **Decipher**, a new database of chromosome anomalies identified through genomic array technology and other molecular cytogenetic methods

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TREASURER'S STATEMENT

Income

We have been particularly thankful to receive a number of major grants this year.

- We are grateful for the second year of The Health Foundation grant which covers the salary of our Information Officer
- We were also successful in receiving the Children in Need grant to cover the Assistant Information Officer's role for a two year period
- We were also very pleased to receive further funding from Garfield Weston (£75,000 at £25k over three years)
- VICTA have supported us for another year with a one year grant of £6,013
- Our application to Lloyds TSB Foundation to cover the part time salary of our new treasurer was also successful this year
- The London Law Trust for £2,000 to cover printing costs of our new information leaflets
- We also received funding from the EC in respect of the two projects we are involved in SAFE and EUROAGENTEST

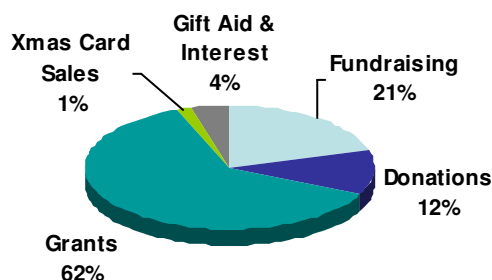
Fundraising activity has resulted in a fantastic year for us.

- Our runners this year entered numerous events including
 - London Marathon
 - Great North Run
 - Blaydon Races
 - Dublin Marathon
 - Nike London 10km walk
 - Caledonian Challenge
 - Half Iron Man
- For the first year we decided to sell *Unique* Christmas Cards, which was very successful not only for raising funds but also from the generosity of members giving extra donations, which resulted in over £3,000

We still rely heavily on members giving donations and we are grateful to everyone who made a contribution no matter how large or small. We are particularly grateful to some of the larger donors including:

- Rolls Royce, Bristol for a £5,000 donation as the result of winning an international competition and nominating *Unique* to receive the award.

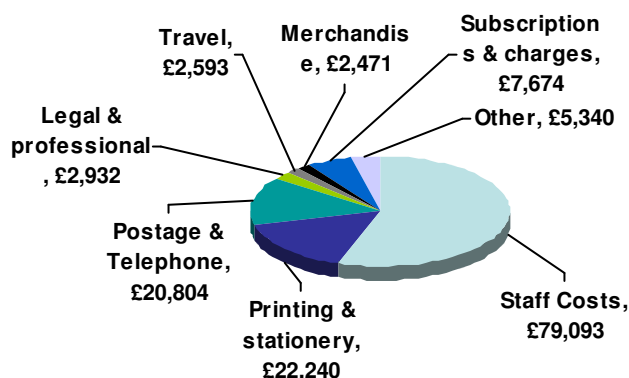
- Shell UK and The Bernard Sunley Foundation for their donations of £2,000
- Bristol & West Plc for a £3000 donation to sponsor the Surf2Sunset event in July 2004.



Expenditure

Our expenditure during 2004-2005 was generally against budget.

Since postage and printing costs to cover the three newsletters during the year account for a significant proportion of our expenditure it is noted that costs have increased since 2003, as the newsletter has now increased to a 36 page colour magazine, which has in turn increased postage costs due to its weight.



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Audited statement of financial activities

Incoming Resources	2005 total	2004 total
Grants	134,357	4,550
Donations	30,603	20,662
Fundraising	44,715	34,060
Christmas Card Sales	3,208	-
Bank Interest	3,924	2,960
Total Incoming Resources	216,807	62,232
Resources Expended	2005 total	2004 total
Staff Costs	79,093	70,184
Printing & Stationery	22,240	17,716
Postage & telephone	20,804	17,442
Subscriptions & Charges	7,674	3,656
Equipment & Computer Costs	2,302	5,658
Legal & Professional	2,932	564
Travel & Subsistence	2,593	2,061
Merchandise costs	2,471	-
Project costs	1,253	-
Other Costs	1,447	6,200
Depreciation	338	-
Total Resources Expended	143,147	123,481
Net Movement in Funds	73,660	(61,249)
Funds at 1 April 2004	63,622	124,871
Funds at 31 March 2005	137,282	63,622

Balance Sheet

	2005 total	2004 total
Fixed Assets	1,013	
Debtors	3,158	1,719
Cash at Bank and In Hand	137,399	62,467
	140,557	64,186
Creditors: Amounts Due within 1 Year	4,288	564
Net Current Assets	136,269	63,622
Net Assets	137,282	63,622
Restricted Funds	22,146	-
Unrestricted Funds	115,136	63,622
Total Funds	137,282	63,622

Approved by the trustees on 17 September 2005

E. M. Knight

Edna Knight
Group Co-ordinator

Gillian Manvell
Gillian Manvell
Trustee

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These summarised accounts may not contain sufficient information for a full understanding of the financial affairs of the charity. For further information, the full trustees' annual report and accounts should be consulted. Copies are available on request from the Treasurer: call 0117 979 8886. The full accounts have been filed with the Charity Commission.

We have audited the full annual accounts for the charity for the year ended 31 March 2005; these were approved by the trustees on 17 September 2005. We issued an unqualified opinion on those accounts. In our opinion, the statements shown here are consistent with the full accounts. Godfrey Wilson Ltd, Chartered Accountants & Registered Auditors, 48 West End, Minchinhampton, Gloucestershire GL6 9JA

 29 SEPTEMBER 2005

THANK YOU TO

Like most charities we rely heavily on the generosity of individuals who contribute through giving their professional expertise for no recompense.

We are extremely grateful to Trevor Searle for his expertise, time and patience in improving and maintaining the **Unique** website and online capabilities. Without this in-house resource **Unique** would not be able to support the financial commitment needed to support our website.

We would also like to extend our thanks and appreciation to Carey Hunt, who provides us with her creative skills to help produce such professional looking literature including the newsletter.

We would not have managed through the legal minefield of paperwork without the support of Nigel Barrett and Gillian Manvell who have worked closely with the solicitors KJD to ensure that every member of the committee fully understands the implications and responsibilities which will occur as a result of **Unique** becoming a Limited Company.

Our thanks also go to two new committee members. Eleanor Fiske, who has taken the minutes of two committee meetings, provides legal support and has helped our Information Officer research specific chromosome disorders. Michael Patterson's wealth of knowledge on a range of issues affecting charities has been extremely valuable.

Over the last year Professor Maj Hulten has been a major influence and support in our work with our EU partners.

We would also like to recognise the efforts of our members Brian and Jo Moore who collate the

newsletters, put them in envelopes and mail them on our behalf.

There are a number of specific fundraisers we wish to thank including:

- Vanessa Roberts for raising over £4,300 from an organised Bollywood event
- John and Myra Knight raised over £1,000 from their Christmas Lights display in Bedfordshire
- Doreen Baxter for selling her hand made cards
- Julie Griffin and Sarah Trevitt for organising the Surf2Sunset event in July which raised £11,000 between Unique and Springboard Opportunity Group, Clevedon
- All our runners: Nick Wellsted, Paul Mannard, Christine Murray, Graeme Tones, Zena Sheen, Diedre & Gareth Edwards, Alyson & Richard Weymes, Bianca Roccelli, Andrew Lister, Peter Hubbard, Justine & Keth Baird
- Sioux & Alan Clarke for their P&O Oriana marathon
- Tina Branden Whitaker for putting on a concert in aid of **Unique** in Brixton in December 2004
- Lucy Austin and Andrew Tickle for all their fundraising over the years

Unique would like to say a generous thank you to everyone who has raised funds for us, from £5 to £5,000. Every penny is put to good use and we couldn't do it without your support.

Lastly, although by no means least, we thank Edna Knight our Group Founder for her inspiration and commitment to the group over another year. It is a great achievement to her and the team that we are able to celebrate 21 years supporting **Unique** families.

Let's hope that we can continue to find the funding to remain active over another 21 years.