Unique™ Conference 2008

Overview

From Friday 31st October to Sunday 2nd November 2008, almost 400 people came together in Daventry, UK for *Unique's* 12th family conference. Families, friends, geneticists, counsellors, doctors, psychologists, scientists and behavioural and educational experts came from round the world to join *Unique* members: from the USA, Australia, Ireland, Norway and Sweden as well as the UK's constituent countries, Scotland, Wales, Northern Ireland and England.

On Friday evening, after a welcome from Gene, *Unique's* new mascot, and from entertainer and magic man Peter McKenna, families who had requested a session with a clinical geneticist were able to talk privately in 1:1 clinics with one of the remarkable team of professionals who so generously gave their time and expertise: Dr Simon Holden from Cambridge & London, Dr Alex Magee from Belfast and Dr Sue Price from Northampton, joined on Saturday by Dr John Tolmie from Glasgow and Dr Helen Cox from Birmingham.

On Saturday morning, having left their children in the crèche, delegates gathered to listen to two inspiring presentations on positive ways forward with rare chromosome disorders (see page ii).

In the afternoon, families gathered in smaller groups for workshops – on challenging behaviour, communication, chromosomes in general, chromosome translocations and sex chromosomes as well as the experience of living with an affected child. Workshop leaders brought a personal or professional lifetime's expertise to their sessions: on challenging behaviour, Chris Oliver as Professor of Neurodevelopmental Disorders at the University of Birmingham and Director of the Cerebra Centre for Neurodevelopmental Disorders, ably assisted by trainee clinical psychologists Ruth Fishwick, Abby Marr and Dr Fay Cook; on communication, Emma Barker as a speech and language therapist now in independent practice; on sex chromosome aneuploidy, Victoria Leggett, the research assistant for the Sex Chromosome Aneuploidy study now under way at Oxford University, as well as clinical geneticist Dr Debbie Shears and genetic counsellor Vishakha Tripathi; on chromosomes in general, Drs Cox, Magee, Price and Tolmie; and on chromosome translocations, Dr Holden and *Unique's* medical adviser Professor Maj Hultén. In addition, an amazing team of cytogeneticists and molecular geneticists led by Val Davison, director of the West Midlands regional genetics laboratory, gave parents hands-on experience of looking at and analysing chromosomes. Meanwhile experienced parents Liz Barraclough, Marion Mitchell, Marie Layng and Patricia Tandy shared their lived experience of having a child with a rare chromosome disorder in the family.

You can find reports of varying length on the workshops on page v. The chromosome/DNA analysis display is reported on page iii.

At tea time, families and children came together to round off the day and to hear *Unique* finance officer Julie Griffin talk about the group's finances. She told delegates that the conference raffle had raised £265 and that her son Nathan had used his budding salesman talents to raise £89 from selling lunchboxes, Tracker bars and cuddly penguins generously donated by *Unique* member Laura Sherwood-King. Fundraising makes up a vital 47 per cent of *Unique's* income, Julie told delegates. Fundraising highlights of the past year included £8,500 from the Gatwick Airport Pantomime Society, the Great British Duck Race, the Bristol Dragon Boat Race which raised over £4,000 as well as running events such as the Great North Run and the London Marathon. Julie stressed that small, local events are vital too – coffee mornings, birthday party donations, games and quiz nights all help to keep *Unique* afloat. In 2009, *Unique* will be 25 years old – can you do something different to celebrate this?

Early in the evening, the geneticists were available again, seeing families for a private 1:1 consultation while other families took their pick from an array of therapists offering private consultations.

With the main conference over at just past five o'clock in the afternoon, you would have thought the delegates would be tired but it was just the opposite! This time around, we wanted to offer the delegates a chance to relax and unwind with some exciting and well known therapies. We arranged for four fully qualified holistic therapists to pamper delegates with a choice of holistic treatments (reiki, reflexology), massages and beauty treatments. The appointment lists filled up quickly and by midday there were only a couple of free slots. The therapists created a calm and tranquil setting in the rooms with lovely scented candles, heavenly massage oils, dim lights and, of course, soothing music. It was wonderful to hear that everyone enjoyed the sessions. The therapists were equally appreciative. They were happy to be involved in the conference and full of admiration for the families that they met, and are already looking forward to the next *Unique* event!

And so the family get together began in the evening. With everyone spruced up and nourished, magic/discoman Peter McKenna started off the disco. At 8pm we had a performance by the Rhythm Blasters Dhol Group. The dhol is a double-sided barrel drum played mostly as an accompanying instrument to the traditional Punjabi dance of Bhangra. People who play Dhol are called 'Dhol Players' or 'Dholis'. The drum is played using two wooden sticks, usually made out of bamboo and cane wood. Thank you to Bobby who sent three of his lads to entertain us. They were led by Kinda and performed an exhilarating routine. Then – oh yes, there was more – we had two performances by the Apna Gidha Group and an appearance by Gene. Gene didn't want to be left behind; he enjoyed the disco and enthralled the children. At times it was like watching the Pied Piper… how the children followed him around and were so excited to see him! The Apna Gidha Group delighted us with two amazing gidha dance routines. Gidha is a form of indigenous Punjabi folk dancing performed by women. (Bhangra is the most commonly known Punjabi folk dance and is primarily performed by men.) Thank you to Jasvir K Chohan who sent us her dance group led by Simmarjit Kaur. They had everyone clapping and moving to the beats and music… and they fascinated everyone with their colourful costumes and swaying hips! But the evening didn't end there. Keith Henry thrilled everyone with his rendition of Michael Jackson's moon walking and there was lots of reminiscing with the 'Macarena'. It was past one in the morning when many made their way back to their rooms – still careening to the music!

Sunday morning, usually wind-down time at *Unique* conferences, was nothing of the sort: a stress-busting laughter workshop led by *Unique* father Raymond Polack, followed by *Unique* member and triple graduate Kathryn McKerracher's presentation of the self advocacy tool she has developed and finally a meeting that decided to launch a new charity for families affected by chromosome 18 disorders. You'll find reports of all these sessions on pages vii–viii.

By mid-morning, it was all over: the magic man's modelling balloons had gone home with the families or burst and hugs and emotional goodbyes signalled that it was time to go.

Unique's 12th family conference simply would not have been possible without the vast number of helpers who gave their time and skills freely and generously to support the group's seven staff. Thank you from the bottom of our hearts, everyone!

Edna Knight, founder and life president – quote

'Unique was started about 25 years ago with five families and an exercise book. It's hard to believe that it's come so far.'

Edna, founder and life president, was awarded an MBE in this year's Queen's Birthday Honours list for voluntary services to people with a chromosome disorder and their families. She will be receiving her award on 10th December at Buckingham Palace. Amy, the 21-year-old daughter of former *Unique* treasurer Nigel Barrett, marked the honour by presenting Edna with champagne and chocolates.

Beverly Searle, chief executive officer – quotes

'We have a lot to celebrate. We now have over 6,300 families in 76 countries, representing over 7,000 individuals with a rare chromosome disorder, many of whom are not written about in the medical literature. This represents a huge resource of information for families and for professionals worldwide. Membership is growing at 800 new families a year. By being able to provide professionals with information to support other families, our reach extends well beyond our database.

'We have produced more than 100 family-friendly leaflets on specific rare chromosome disorders, covering thousands of individual disorders, and these leaflets are freely available to download from our website. We have worked with large numbers of genetics professionals, including the 30 to 40 geneticists here today.

'We are delighted to announce that three eminent geneticists have already agreed to be patrons of Unique: Professor Albert Schinzel from Switzerland; Professor Jean-Pierre Fryns from Belgium; and Professor Judith Hall from Canada – and Professor Dian Donnai from Manchester has also assented.

'We have been granted HonCode certification to show that we comply with the rules that govern providing reliable medical information on the internet.

'Recently we have joined social networking sites: we are on FaceBook and on MySpace; we have a Cause with 700 people supporting us. We now have a new regular electronic news alert.'

Plenary session:

Positive advances in services for disabled children – what the Every Disabled Child Matters campaign has delivered

Srabani Sen set the upbeat tone for the conference. Srabani leads the umbrella group Contact a Family as its chief executive officer and is a board member of the Every Disabled Child Matters campaign – known as EDCM. This is an English campaign to ensure that disabled children and their families enjoy the rights and justice that are due to them. It's a joint initiative between four groups – Contact a Family, the Council for Disabled Children, Mencap and the Special Education Consortium – who came together when they realised that together they were more likely to be heard than when they made a noise individually.

'We recognised that disabled children and their families figured nowhere,' Srabani said. 'They were nowhere on anyone's agenda, politically or locally. Nobody was bothering about the issues that affected them. We also recognised that if we were going to make a difference, we needed investment, we needed money.'

Huge increase in families with a disabled child

The number of children with a severe disability is growing: every 25 minutes another child is diagnosed. In the 27 years to 2002, the number of disabled children in the UK rose 62 per cent by 294,000 to a total today of more than three-quarters of a million. Growing numbers of these children have complex needs. What is more, their families are more likely to live in relative poverty, partly because the mothers of disabled children are much less likely to have a paid job. While 16 per cent of mothers of disabled children work outside the home, 61 per cent of other mothers do. A consequence of this is that the most common reason for families to call the Contact a Family helpline is because they are struggling with money. A Contact a Family survey published in September 2008 revealed that one family in six is now struggling to pay for the most basic amenity – food. One family in 14 is at risk of losing their home. Almost half have borrowed money from friends and family and one in five of them is using it to pay their heating bills.

Financial strain and fighting for the services that are theirs by right adds to the stresses that families with a disabled child already face. One third of families report relationship stress; 13 per cent say they have major problems and one family in 11 breaks apart.

Patchy services

Across the UK parents can receive a very varied level of services. A report last year told the stark truth: 'Services remain variable and in some areas very limited and insufficiently joined up. Parents… are dissatisfied with the overall experience. Despite the principles of the England-wide campaign for children Every Child Matters, they feel there is a lack of information, too much duplication of assessments and services, access is difficult and there are high thresholds to social care support. Parents and young people say they do not get enough respite and what is provided is often inappropriate.'

What do you want instead? What does your child want?

So EDCM asked disabled children and young people for their wish-lists: if they could change one thing, what would it be? Number one for youngsters was things to do and places to go; number two was to be respected and number three was a good education. Parents' number one wish was not to have to fight for support; number two was to be included in their communities; and number three was appropriate educational provision for their child's needs.

Short breaks

So what has EDCM achieved? How far has it got?

The jewel in EDCM's crown is that from 2011 families with disabled children will have a statutory right to short breaks. This is spelled out in Aiming High for Disabled Children, an England-wide strategy to transform the lives of families. Critically, it is backed with substantial sums of money – £370 millions for short breaks; £35 millions for childcare; £19 millions to support the programme helping disabled children make the transition to adult services; and £5 millions to enable parents to be consulted about the new services. Sadly, in Scotland, Wales and Northern Ireland no money has been set aside. But lobbying in the devolved states goes on.

Other steps

Meanwhile, 93 per cent of families with a disabled child say they face financial difficulty because of their extra costs and low income. Yet a possible 100,000 people entitled to the Disability Living Allowance are not claiming it. As a result of lobbying, the government is

promoting the allowance – which can make the difference between being able to feed your family and not. EDCM is also targeting a winter fuel allowance for families with a disabled child. In Scotland, the Scottish Executive has recognised that families with disabled children are at risk of fuel poverty and will be putting forward proposals to tackle it. Unfortunately, there is no such recognition in England – even though it would only cost the government £2.5 millions – peanuts – to give a winter fuel allowance to the poorest 12,700 families with a disabled child. Having established that disabled children may be 'the worst housed group in Britain', EDCM is also targeting more funding for adaptations, better regional planning and for all disabled children to have their own bedroom.

What next?

EDCM has made disabled children a national priority for this government. But how long will this government last? And how sincere are the Opposition's 'warm words' of support?

The decisions that are taken locally are the decisions that impact on families, so EDCM is working locally to ensure that local authorities and primary care trusts (PCTs) make disabled children a priority. The commitment from local authorities has been greater than from PCTs.

'So – has EDCM improved things for disabled children?' Srabani asked. 'Yes, but…' In England they are much higher up the political agenda; in Scotland there is some progress; in Wales lobbying has been much harder and not much is happening in Northern Ireland.

'Whether we genuinely achieve the government's ambitions around transforming the lives of children with disabilities is a question mark.

There is still a long way to go,' Srabani concluded.

Families then asked Srabani questions.

Question: What is a short break?

Answer: *The government says the definition should be as flexible as what families say they want. For some it'll be a break away. For others it'll be for someone to come in and look after their children. That's why parental input into designing the service is so important.*

Question: Short breaks are never mentioned locally in my carers' groups in Birmingham.

Answer: *They should be. It's critical that you go back and ask why you have heard nothing about them. The responsibility lies with local authorities and PCTs to tell you about the scheme and about how you can contribute to making decisions locally. If there is a gap between what the government thinks is happening locally and what is really happening, we need to know about it. From my position on the Ministerial Implementation Group, I can then tell ministers what's really happening.*

Question: We understand in Shropshire that the joint funding for short breaks that local authorities have been told to expect from PCTs may not materialise. We may get some money but it won't be equal.

Answer: *Last year's Comprehensive Spending Review specified what money the government is putting into local authorities; the government said they were putting an equivalent amount into PCTs. Since then we have repeatedly asked the government to specify how much money it has given PCTs but they have yet to say how much it is. Families must keep asking at their end and EDCM will keep asking at theirs.*

Plenary session:

Positive Perspectives on Rare Chromosome Disorders

Dr Linda Gilmore, an educational psychologist from the Queensland University of Technology, Australia drew rapturous applause and warm support with her talk on new and positive ways of approaching rare chromosome disorders.

Already interested in disability and Down's syndrome, Linda's first brush with rare chromosome disorders occurred when she met a family with a one-year-old daughter with an 8p deletion. The family had just received the diagnosis and had been told – quite incorrectly, as it turned out – to expect the worst: a maximum lifespan of 12 years and a vegetable-like existence. As Linda looked at the little baby, she appeared quite normal to her. She resolved to find out more for the family.

'I was appalled that I couldn't find any information,' Linda told delegates. 'First it took me a long time to figure out what having a deletion on a chromosome meant. Then what bothered me was that I couldn't find any literature. There was nothing to tell me how I could work with this family whom I wanted to work with and support. I didn't know what the implications were. The limited literature that I found was so grim.'

But Linda did work with the family and she found that this child has normal intelligence. 'I was startled to discover that the literature was wrong and gave the wrong impression. I subsequently worked out that the literature is overly negative,' she said.

Negative bias in medical literature

This experience alerted Linda to the strong negative bias in reports on rare chromosome disorders in the published medical literature. There are many things wrong with the reported literature, Linda said. Overwhelmingly, it's about medical and genetic factors. The bias towards negative cases runs right through, due in part to the fact that children with more serious consequences of their chromosome anomaly are more easily recognised and diagnosed and so reported. Even editors of professional journals tend to publish cases that are more serious and are less interested in publishing cases where everything is going reasonably well. Where a child is developing typically, that tends not to get reported. Reports are also made at one point in time, yet families know from their experience of their own children that they are not the same this year as next. There is little consideration of the family context and other things going on around the child.

Linda also discovered that there was often very limited research about rare chromosome disorders, leaving many families with a child with a karyotype there simply is no published literature about. At most there are isolated case reports because there are usually not enough people with a particular karyotype to analyse group data and draw any conclusions.

Overall and most importantly, there is inadequate information about the often wide range of possible developmental outcomes for children with rare chromosome disorders. There is very little of what families really want to know – whether this is a child who will walk and communicate with her friends and go to school. Instead there are case reports that tend to be impersonal and give little sense of the person.

One result of the generally inadequate literature is a lack of knowledge among professionals about chromosome disorders – teachers for instance asking for a 'name for the condition rather than the karyotype and asking whether the child would grow out of the condition'.

Even more seriously, professionals base their prognoses – the predictions they give families – on published cases. Families are encouraged not to raise their expectations. When a condition is rare, it's inevitable that there will be uncertainty when the diagnosis is given but, Linda said, that gives grounds for cautious hope. 'If the outcome is unknown you can be guardedly optimistic about how things will go.'

Inadequate support and family isolation

On top of this there is inadequate support and family isolation. 'I constantly meet families who have had a diagnosis from a geneticist, seen a

paediatrician but not been told about groups like *Unique*.'

Professionals she has found focus too often on problems, weaknesses and deficits – because they think these are the things that concern families and that they therefore need to do something about.

In what areas is a child developing typically?

'We focus on weaknesses at the expense of focusing on things where the child is developing quite normally and typically. We don't take enough notice of the areas where the child has strengths. Yet for anyone it's very important to nurture strengths.'

Because there is not enough recognition of a child's potential strengths, there is uncertainty about appropriate interventions. What is more, the failure to focus on strengths feeds perceptions of disability that are often unnecessarily negative and pessimistic.

Resilience and positive psychology

By contrast in psychology for the past 20 to 30 years there has been a more positive focus on resilience – examining how it is that some people do better than others when faced with adversity. Instead of studying people with weaknesses, psychologists have been looking at why some children do well despite growing up in adverse conditions. A more recent trend is 'positive psychology' – looking at what contributes to happiness and to fulfilling, meaningful lives. As they try to understand positive states such as satisfaction and what it means to be human, psychologists are shifting away from pathology. Yet this huge and growing field of exploration hasn't filtered through to disability.

But what do psychologists know about resilience in the families of children with disabilities? About families they know a little. A generation ago there was a presumption that having a child born with a disability was negative, leading to grieving and chronic sorrow. More recently, the focus has shifted to the fact that the families may be stressed but they cope as well as they can, some better than others. The concern is then how one can work with families who are not coping so well? Even more recently, people have called for us to focus on the positive aspects of having a child with a disability born into a family. The point here is that a lot of families have reported that having a child with a disability is really a positive thing in their lives that has made their lives more valuable and helped them to lead richer lives and to reflect on what is really important. Families learn to take things one at a time. They say the experience has helped them to slow down and appreciate things more. Not only parents are affected: brothers and sisters too report greater tolerance, more empathy and a greater sense of social justice and loyalty – all positive benefits that would not have come about in the same way had the disability not occurred.

Linda conceded that not all families experience these positive things and they don't experience positive things all the time. But she pointed out forcefully that the positive aspects need to be taken into account. 'We need a balanced perspective on families of children with disabilities,' she said. 'We need to recognise that while at times things are incredibly stressful, for some families some of the time there are tremendous positive gains.'

Linda explained that she is currently working on building in resilience factors for children making the transition from primary to secondary school. 'We are trying to establish the networks of social support that we know are tremendously important to typically developing children; we are working on children's skills in speaking out and standing up for themselves; on dealing with bullying; dealing with being different; identifying feelings and expressing them.'

Grounds for cautious optimism

There are reasons, she said, to have expectations that are cautiously optimistic. Some children do develop better than others. Some children with a range of chromosome abnormalities do have normal intelligence – not a huge number, but the literature is biased to the negative. She encouraged families to get their professionals to help them to understand the published reports in the medical literature and not to take them at face value.

Linda explained that positive perspectives are also about thinking differently. She recalled the Welcome to Holland account of the experience of discovering that you have a child with a disability, where a family expecting to enjoy a holiday in Italy arrives in Holland instead. They find that Holland has a lot of things to enjoy – tulips and Rembrandt. Linda suggested that some people end up in Turkey or another country. 'The pain of missing out on Italy doesn't go away because you've lost that dream, yet you have something else,' she said.

She explained 'reframing', where psychologists try to get people to see things in a different way, and recalled a support group for the parents of children with extremely difficult behaviours who ran a weekly competition for the worst behaviour. Competing for the weekly 'worst behaviour' prize did not cure the problem, but it broke the stress of dealing with it and perhaps made it a little easier to deal with. Linda reminded families that the things they remember long-term may be the positive aspects. 'It's important to try to get things in perspective. If you can support each other to do that, it can be tremendously valuable,' she said.

Positives about disability

She said that despite the challenges and difficulties there are positives about disability. 'There have been times when I have looked at someone with an intellectual impairment with a sense of envy that they live more in the moment and their lives aren't cluttered up with paying next week's bills and other demands and lacking that sort of clutter gives them the ability to enjoy life in a purer sense.'

She admitted that she finds people with disabilities more interesting than 'normal' people and reminded the conference that people with disabilities make up at least 20 per cent of the population. 'We are all different. Can any of you claim to be really, really normal?' she asked.

She pointed out that there are many things we can do to promote understanding of disability. She showed a card from the National Autistic Society designed to be handed out when a young person with autism is being disruptive or noisy in public. The card reads: This young person has autism. Autism is a developmental disability that affects social and communication skills. People with autism may behave in unpredictable ways as a result of their difficulty with understanding language and social situations. Please help us by being understanding and showing tolerance.

She highlighted the importance of portraying people with disabilities positively and the work of Rick Guidotti **(www.positiveexposure.org)**.

Finally, Linda concluded by reminding the conference that in many ways children with disabilities are just like other children. 'The disability is not what defines a person; it's a little bit of them. It's really important to remember that and to stress that to the professionals that you work with.'

Question: How do you explain the disability to siblings, particularly those younger than the disabled child?

Answer: *It depends on the age of the children and how much they understand of what is happening. Unique Tales can be a fantastic way. It's never too early to get across the message that we are all different. We all have things that we do well and things that we don't do very well. I talk to children sometimes about the wiring in the brain; some wires are stronger than others and some parts of the brain are not as well developed as others. So some children have more of those loose connexions. I always look for something that the child with a disability can do that the typically-developing siblings can't do so well. If you look hard, you'll find something.*

It can be difficult if you have a child with a lot of behaviour problems and you have a brother or sister who's embarrassed or finding it difficult to cope with. Sometimes brothers and sisters have difficulty in explaining to their friends about their brother or sister who has a disability. Stress the idea of being different and showing some understanding.

Workshop: Challenging behaviour

Professor Chris Oliver and his team, Department of Psychology, University of Birmingham

The challenging behaviour workshop was the most popular of all the workshops suggesting that behavioural issues are a concern for many parents. Chris Oliver was keen to stress that in many ways children and adults with a chromosome disorder are just the same as everyone else: they have more in common with other people than they have differences. Both biology and the environment shape and effect how we behave. Indeed, the coping techniques that he outlined are relevant to all parents and carers! However, he pointed out that it is important to acknowledge that the chance of certain behaviours does increase in some syndromes or chromosome disorders. He and his team are interested in the reasons for these behaviours. A greater understanding of how and why these behaviours occur will hopefully guide more effective treatment and education. Chris Oliver used his extensive years of experience and research to deliver an engaging and interesting talk full of ideas and management strategies.

He began by discussing the role of genetics in behaviour. By and large people acknowledge the idea that genetics plays a part in how we look. However, people tend to be less comfortable with the impact of genetics on behaviour. Nonetheless, it is clear that genetics has a central role in some behaviours. Chris Oliver used an example, Angelman syndrome (a genetic syndrome in which a set of genes on chromosome 15 are inactivated) to illustrate this point. A person's genes are the code that specify how the body is to grow, develop and function. Children and adults with Angelman syndrome have gait ataxia – they walk with their legs widespread and their arms held up. They often have jerky movements. This tends to be classified as a behaviour but can be explained by genetics. One gene affected in Angelman syndrome, UBE3A, is expressed in the brain within the region that controls those actions that we perform routinely without specifically or consciously thinking about them, such as walking. If the code is not given correctly as in the case of Angelman's syndrome, the ability to walk naturally without giving it specific thought is lost. Therefore, the brain compensates and the children and adults can and do walk. However the action of walking is no longer an activity that is performed unconsciously without active thought and results in the pronounced, jerky movements.

The impact of health and pain can also have a significant effect on behaviour. Chris Oliver illustrated how pain can lead to challenging behaviours using Cornelia de Lange syndrome (CdLS; a rare genetic disorder that affects both physical and intellectual development) as an example. Almost all of those with CdLS are affected by gastro-oesophageal (GO) reflux (feeds and stomach acid return readily up the food pipe from the stomach) causing pain and discomfort which can manifest in behavioural associations. The pain that the GO reflux causes and attempts to allay this pain can result in fidgeting, hands or fingers in the mouth, excessive salivating, an arched back or grinding of teeth. Children with GO reflux may also be more likely to self-injure (cause physical discomfort to themselves) and the pain may be an important indication of why they do so. The pain caused by the reflux can be blocked (gated) by rubbing, scratching or hitting another part of the body. However, there are often sensory differences associated with chromosome disorders resulting in signals carrying the blocking signal working more slowly so that the child rubs, scratches or hits more and harder which can then lead to chronic pain and so the loop may begin again. Chris Oliver described a boy with CdLS suffering from GO reflux together with self-injurious behaviours. He underwent a fundoplication (a surgical operation to improve the valve action between the stomach and food passage) which removed the pain and the self-injurious behaviour ceased.

Chris Oliver then went on to talk about repetitive behaviours which sometimes affect children with chromosome disorders, and suggested strategies that may help to manage them. The key points to remember when dealing with repetitive behaviours:

- Aim to modify (reduce or restrict) the behaviour and not eliminate it
- Aim to make gradual ("graded") changes rather than sudden changes
- Bear in mind that it may take a long time before you see results.
- Deal with one behaviour at a time
- In young children, prevention works better than intervention so keep an eye out for any new "behaviours" and deal with them promptly.

Communication problems can also lead to challenging behaviour. All children need a way to communicate their needs and children with a chromosome disorder accompanied by a communication difficulty may have less means at their disposal to get something that they need or want (such as food, toy or a preferred activity) so they resort to challenging behaviour in order to get it. This can very easily become a behavioural loop which can be hard to break. The child has a need for others to do or give something and so uses some form of challenging behaviour to make the point causing frustration, confusion or distress which is acted on (by reprimanding, comforting, distracting) which can act as a positive enforcement or reward. The fact that the challenging behaviour has worked and got a response increases the chance of challenging behaviour occurring in the future. Although Chris Oliver acknowledged that it is easier said than done, he advised: be aware of how you respond. Are you rewarding problem behaviour?

He summarised this interesting and rewarding session by saying that social contact is a potent reward for both desirable and undesirable behaviour – use it wisely! Challenging behaviour has a strong learned component, therefore behavioural management can help.

Chris Oliver and his team have produced a book entitled *Self Injurious Behaviour in Cornelia de Lange Syndrome: A guide for Parents and Carers*. Much of the content is relevant to children with genetic disorders other than CdLS and is available directly from Chris Oliver. Please email him on **c.oliver@Bham.ac.uk** if you would like a copy.

Workshop: Communication

Emma Barker, an independent speech and language therapist, presented the workshop on communication

She opened by pointing to the multi-faceted nature of communication and how understanding and expression, visual and aural information and memory are interlinked.

She showed a five-tier communication pyramid. Tier One, the foundation, comprises attention and listening. For any kind of language development, children need to pay attention – even for just two seconds. She stressed how important it is to make the most of the child's attention span. Tier Two in the pyramid is play and interaction – setting up opportunities for children to look at things and communication opportunities, giving children opportunities to play and explore and learn through practical experience. Tier Three is receptive language (understanding), which is much better in many children than expression. Emma pointed out that some parents focus so completely on encouraging their child to speak that they overlook the need to develop their child's understanding. Tier Four – expressive language – can use any mode of communication including facial expression and high and low tech communication aids. Tier Five – the icing on the cake – is speech and pronunciation. 'The most important thing is functional communication and getting your message across. Can you get your wants and needs met? Can you comment about things? The fact that you can't say 'k' isn't a priority in the grand scheme of things.' All the same, if people use speech to communicate, then intelligibility is important. If this is a stumbling block, can you use other ways to help a child to get their message across? Emma stressed that if you are working on speech, motivating children and making the job fun and interesting really matter.

Alternative and Augmentative Communication (AAC)

AAC describes different ways of helping people with communication difficulties to communicate. A communication aid can be as simple as a piece of cardboard or a complex computer-based system. It can supplement speech, it can turn the child's way of communicating into speech or it can facilitate speech. Parents often ask whether using a sign system such as Makaton will stop their child from speaking. All the research and Emma's experience backs the view that it's important to see the extra communication system as a facilitator of communication rather than an end in itself. Some children will sign into adulthood. Some children use signs alongside words. Some drop signing as soon as they can say words. 'There is no ideal form of communication aid. Choose what's appropriate for your child in whatever context they find themselves.'

Children need to want to communicate and parents may need to create opportunities. For some children communication isn't natural and they'll do everything to avoid it. Once a child has learned to use a communication aid, it's important to always have it available – otherwise they are rendered voiceless.

Low tech aids

These are anything that is non-electronic and include symbol systems, objects of reference, words and line drawings. They are largely ways of presenting concepts visually and while there are many ready-made systems – Makaton, Boardmaker and PECS images are just some – there is no reason why families can't use their own images for example by downloading Google images off the internet. Symbols can then be used in various ways – to express need and wants, as in picture exchange systems; to track time as in diaries; to tell stories; to offer choices; for pointing activities. They can be built up into communication books and boards, exemplified by an extensive communication board developed by Wendy Marriott for her adult daughter Rebecca and on display during the conference. They can be used in tandem to reinforce messages, for example alongside signing. Some children can acquire phenomenal numbers of signs which they can use to chat with, to relate events from the past or plans for the future.

High tech communication aids

These aids are usually electronic and range from devices that rely on simple choices to complex devices that offer speech output. Parents are often desperate for their child to use high tech aids, Emma said. But she cautioned that a lot of the foundation work is done with low tech aids before a child is able to go on to use a high tech aid efficiently. Families also need a lot of support in setting a high tech aid up, maintaining it and adding new vocabulary. Families should think carefully about the physical limitations of some devices – they may be bigger than the child; they may not be weather-resistant – and about who will communicate with the child using the particular aid. They should make sure that their child has a really thorough assessment and that they are properly taught how to use the aid and are ready to develop it.

Two families with adult daughters then demonstrated the aids they use: Julie Bello and her daughter Natasha, who uses a high-tech voice output device and Wendy Marriott and her daughter who uses an extensive communication board.

Two charities that support augmentative communication users and their families are Communication Matters **(www.communicationmatters.org.uk)** and I voice **(www.I voice.info)**.

Contact details for Emma Barker are **mail@thechildrenspractice.co.uk**. The website is at **www.thechildrenspractice.co.uk**.

Conference Lost and Found

Left in hotel during the weekend – young girl's black jacket. Please contact Marion if it is your daughter's.

Tel: +44 (0) 1293 525504
Email: marion@rarechromo.org

Workshop: Experienced Parents

This year, for the first time at a *Unique* conference, a number of 'experienced' parents ran a workshop. Four *Unique* parents agreed to share their experiences of parenting children with a rare chromosome disorder and offer any advice or pearls of wisdom that they may have gained along the way! Liz Barraclough, mum to Eleanor age 13 who has an 8p inverted duplication, kicked off the session. She began by introducing Eleanor by showing a picture of her karyotype to demonstrate how Eleanor is often treated by professionals as a karyotype and how important it is to remember that Eleanor is her beautiful little girl. She was joined by Marie Layng, one of *Unique*'s committee members and mum to Dominc, now 22 years old who has duplication of 2p; Marion Mitchell, *Unique*'s Family Support Officer, organiser of the *Unique* conference and mum to Rob, 14 years, who has Idic 15 and Patricia Tandy from Australia whose daughter, Laura is 28 and has 1p36 deletion syndrome. Together they shared their stories and invited other parents to contribute their experiences. In this lively and rewarding session, parents were able to ask questions and seek advice from other parents who had perhaps travelled a bit further down the road or had experienced similar issues. The topics covered were varied and wide-ranging encompassing schooling (both inclusion in mainstream schools and special educational establishments); the individual needs of each child; respite care (which not only gives the parents a break but also gives children a break from their parents!); social workers (it seemed only a minority of families had one); the transition from childhood to adult services and the vast differences between provisions in different areas of the country. It was abundantly clear that the room was over-flowing with a wealth of knowledge and wisdom serving to demonstrate that other parents are often the best source of information!

Workshop: Sex Chromosome Aneuploidies

This workshop was presented by: Victoria Leggett, Department of Experimental Psychology, Oxford, Dr Debbie Shears, Clinical Geneticist and Vishakha Tripathi, Genetic Counsellor

Victoria Leggett is a Graduate Researcher and is part of a team of investigators who are carrying out a preliminary study which aims to provide more complete information for parents who are given a prenatal diagnosis of a Sex Chromosome Trisomy. They have carried out two study days, in collaboration with *Unique*, one for families with daughters who have XXX and one for families with a son with XYY.

Dr Debbie Shears gave an excellent overview of the basic chromosome structure and how sex chromosome aneuploidies come about and what they look like under the microscope. She also described the rarer forms of sex chromosome aneuploidy such as XXYY, XXXX and pentasomy X.

Victoria then proceeded to give an overview of the Sex Chromosome Trisomy study to date and what will be done in the next few months with a view to completion in June 2009. So far parents have given information on 10 boys with XXY, 35 boys with XYY, 33 girls with XXX and their brothers (23) and sisters (9) without a chromosome disorder. The study is considering school performance, attention, social skills and behaviours. They are focusing both on the variability of difficulties seen and their severity to create a profile of strengths and weaknesses.

If you would like to see a copy of the study report so far, please contact Satnam **(satnam@rarechromo.org)**.

During the question and answer session, it became quite apparent that the families welcome any new information/reports that will help them understand their children better and get extra support to help them. The information available in the medical literature is very old and biased.

Workshops: Translocation and Chromosomes in General

Fascinating and wide-ranging discussions were held in the workshops on chromosomes and translocations. Topics raised by families and ably fielded by clinical geneticists Drs Helen Cox, Alex Magee, Sue Price and John Tolmie in the general chromosome workshop ranged from myelination and the recent discovery of the gene behind Pitt-Hopkins syndrome on the long arm of chromosome 18 to the vital importance of tracking the natural history of rare chromosome disorders – a task that *Unique* is well-placed to accomplish. Topics addressed with characteristic verve and humour – as well as underlying serious informational intent – by Professor Maj Hultén and Dr Simon Holden in the translocations workshop ranged from preimplantation genetic diagnosis and the possibility of testing in pregnancy for paternally inherited RCDs with a blood test to the chromosomes of gorillas.

Workshop: Self Advocacy success with 2noMe

Kathryn McKerracher BSc (Hons) MSc MRes presented a tool she has developed for self advocacy. Despite the challenges of deafness, mobility, social and behavioural problems which result from her 18q deletion, Kathryn has not only been awarded three degrees (two in genetics and one in bioinformatics) but has spoken at international conferences and until recently worked at the MRC Virology unit attached to the University of Glasgow. How has she done it?

With the help of self-advocacy. Self-advocacy is fighting for what is due to you, including support; standing on your own two feet; telling people what you need; sharing problems and taking responsibility for your own life.

The challenge

At primary and high school, Kathryn lacked support and this led directly to frustration and challenging behaviours. When she got to college, she didn't know how to ask for support so she didn't – with the result that she nearly failed her first course. Eventually a lecturer reported her problems to the college's special needs department and Kathryn was assigned a Communication Support Worker to take notes, interpret and help her interact with her fellow students. This support continued throughout Kathryn's Higher National Diploma in biomedical sciences. But still she had not learned to ask herself for the support she needed.

When Kathryn got to university in 2000, she depended on the Special Needs Department to assess her support needs. In her first year, she had little help other than from a personal lab demonstrator. In her second year she had an external note taker. Finally, struggling, she approached a lecturer and outlined the problems she was having in tutorials. Together they figured out the best ways for her to get equal access. This was Kathryn's first experience of self advocacy. It worked – and she passed the course.

In 2001, Kathryn was awarded a National Deaf Children's Society award funded by the Millennium Commission and designed to help individuals fulfil a personal dream or challenge as well as benefiting others in the community. Using the funding, Kathryn investigated what services are available to deaf students with additional needs in further and higher educational institutions in the UK and the USA. She visited 13 universities and colleges and found a bewildering patchwork of provision.

A possible solution

But she also found that at the Rochester Institute of Technology (RIT) in New York state, students from the National Technical Institute for the Deaf with whom they share a campus could attend RIT courses with a form that identified their extra needs. Students carried their forms with them and handed them to their RIT lecturers. These forms are the basis for Kathryn's 2noMe.

Back at university in the UK, Kathryn identified and wrote down all the challenges she expected to face and possible strategies to overcome them. She briefly outlined her disabilities – and so the 2noMe was born. 'It is a stress-free method of introducing yourself to others,' says Kathryn.

The solution worked!

Using her 2noMe, the last two years of Kathryn's undergraduate degree course were much easier. She was granted a note taker, some lecturers provided transcripts of their lectures, labs were made more accessible and her tutorials were more deaf aware. Most importantly, Kathryn passed her degree.

Other people who have used Kathryn's 2noMe template have added a photograph and a full list of their difficulties. 'Each document is unique

to yourself, giving you peace of mind in a variety of different situations.'

A 2noMe has been used to help in many different situations – even tricky airport transfers. The benefits are that the individual identifies their own challenges and suggests ways to overcome them, then hands the information over for others to read.

Interested?
If you would like to know more or to see the 2noMe templates that Kathryn has developed to help others to help themselves, please email her at **Ksmckerrachermsc@aol.com**. Kathryn is developing a website at **www.2nome.co.uk** where copies of the templates will be available to download.

Meeting: Chromosome 18 Registry and Research Society

Families affected by chromosome 18 changes resolved at a meeting at the *Unique* conference to set up the Chromosome 18 Registry and Research Society (Europe) as an independent charity. The charity's aims will be to establish a website with information on chromosome 18 disorders and to hold conferences in Europe, the first hopefully to be scheduled for 2009. The new charity will be affiliated both to the Chromosome 18 Registry and Research Society in the USA and to *Unique*, who will send an observer to meetings. A chairman (Chris Wilkinson), a secretary (Bonnie McKerracher) and a treasurer (Linda State) have been chosen and Candy Place has agreed to serve on the committee.

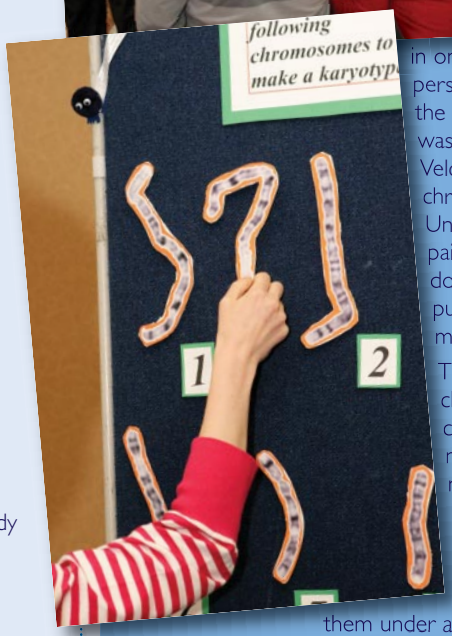
If you would like to join or to know more, please email Bonnie at **bonnie18qmum@btinternet.com**, write to her at **14 Main Street, Twechar G65 9TA, Scotland** or telephone her on **+44 (0)1236 823455**.

Announcement: RISC network

Professor Maj Hultén told families about a project she hopes to launch to improve genetic counselling for couples where one partner has a structural chromosome rearrangement. The project will bring together geneticists, counsellors, *Unique* members and other groups to enable couples to receive more precise and accurate estimates of the likelihood of having an affected pregnancy or baby. If you are interested or would like to know more, please contact Maj at **maj.hulten@warwick.ac.uk**.

Hands-on chromosome/DNA analysis demonstration

Val Davison and her team from West Midlands Regional Genetics Laboratory



The hands-on chromosome analysis room was an extremely interesting demonstration of the process of analysing a person's chromosomes, and while extremely informative, it was also completely accessible and a lot of fun!

There were several display areas, each one dedicated to a particular process of chromosome analysis. The helpful team from West Midlands Regional Genetics Laboratory talked people through each process. First was a demonstration of how by staining a person's chromosomes with chemicals that produce alternative light and dark bands it is possible to visualise the chromosomes under a microscope and distinguish one chromosome from another. Each person usually has 46 chromosomes that come in 23 pairs: one chromosome in each pair from the mother and the other from the father. Examining chromosomes in this way is known as karyotyping and can determine if an individual has a duplication, deletion or other structural rearrangement in one or more of their chromosomes. The summary of a person's chromosome make-up is called a karyotype. Indeed, the highlight and focus of attention for lots of kids (and adults!) was the opportunity to perform a karyotype analysis using giant Velcro chromosomes. One copy of each of the 23 chromosomes was arranged in sequence on the display board. Underneath were a jumble of the 23 matching chromosomes pairs. It was your job to match the pairs – easier said than done! Solving a simplified version of the chromosomal jigsaw puzzle for yourself gave a fantastic insight into the (much more complex) job that the team are doing every day.

The vast majority of *Unique* families will have had their chromosomes analysed in this way. However, very small chromosome changes or rearrangements can be missed or not detected using this technique and this is where the newer molecular techniques come in. One such technique, FISH (fluorescent in situ hybridisation) was on display. It was possible to see via computer technology how FISH uses specific DNA probes to bind to ("paint") small parts of a chromosome in different fluorescent colours and visualise them under a special fluorescent microscope. In this way chromosomes can be examined in greater detail. This is useful if you know which of the 23 chromosomes you want to look at. But what if you don't know where, amongst all the DNA contained within all 23 chromosomes, the change may have taken place? In this circumstance array comparative genomic hybridisation (array-CGH) can be used. This technique utilises a microarray which is, in simple terms, a glass slide on which there are thousands of 'spots' of a reference (control) DNA sample. The control sample is from a person who is known to have two complete copies of each chromosome (as is usual) and is labelled with a colour, such a red. The person of interest's DNA can be labelled with a different colour, such as green, and then be applied to the glass slide. The two sets of DNA bind together and the ratio of red to green in each spot can be analysed to determine if there is the correct amount of DNA, too much DNA (a duplication) or too little DNA (a deletion).

In summary, guided by the whole team, these demonstrations were a wonderful way of helping the non-scientist really start to understand the techniques used by the unit in basic terms and with more detail for the curious-minded.