tac
journal

LEAD ARTICLE - NEW WRITING

The Unique Network: Supporting families challenged by rare chromosome disorders

Beverly Searle

Dr. Beverly Searle: I was appointed to the post of Development Officer for *Unique* in April 1999 and in 2003 I was made Development Director. More recently I became Chief Executive Officer. Before I had my children, I worked for many years as a research scientist, being awarded the degree of PhD for my research into aspects of the biochemistry and genetics of yeast. What a coincidence then that one of my children should be born with a chromosomal defect! When I first had Jenny, I was very upset and really thought that was the end of any decent life for me. Well, even now, it is difficult at times but I can honestly say that in most ways, my life has changed immeasurably for the better and I have met some wonderful people because of Jenny. As for Jenny, despite her disabilities, she is a delightful young lady and her sweet nature shines through. My husband Trevor is a computer software specialist and together we have developed *Unique's* members' database and the group's website.

Summary

In this article Dr. Beverly Searle, who, coincidentally, used to be a research geneticist, introduces IQJ readers to her eighteen-year-old daughter Jenny who was born with a rare chromosomal disorder. She describes some of the great trials and tribulations for families like hers and shows how *Unique*, the organisation she helped develop and now leads, helps families with information and real support.

A brief history of *Unique*

Unique's mission is to support, inform and alleviate the isolation of anyone affected by a rare chromosome disorder. Our fledgling predecessor group was first set up in 1984 by one mum, Edna Knight, who was struggling without support and services to care for four daughters, two of whom were disabled due to a rare chromosome disorder. In desperation, Edna 'found' four other families whose children had the same disorder, Trisomy 9p, and the first *Unique* network was born – a red exercise book posted in turn to each family for them to record their trials, tribulations and tips.

In time, families with other rare chromosome disorders joined the group and in 1993, the charity *Unique* was registered. With the launch of the group's comprehensive database in 1996, followed by the first website and paid employees in 1999, *Unique* has gone from strength to strength. By September 2008, over

6200 families in 76 countries had joined up, with between 800 and 1000 new families currently joining annually.

What are rare chromosome disorders?

We are each normally born with 23 pairs of intact chromosomes in nearly every cell in our body. *Unique's* affected members have extra, missing or rearranged chromosome material which means that all the genes (the instructions that make our bodies work and develop properly) on the involved chromosome segments are also extra, missing or rearranged. Nearly any part of any chromosome or chromosomes can be involved, leading to an almost limitless array of possible disorders. The chromosome abnormality itself cannot be 'cured' but individual symptoms can be dealt with as they arise. While each of our members is quite literally unique, there are common developmental, behavioural, emotional and clinical concerns across our membership.

Developmental delay, growth disorders, a variable degree of intellectual disability, difficulties with feeding, communication difficulties, dystonia and gait problems, sensory disorders and unusual behaviours affect a high proportion of our membership. Clinical concerns are also common and, depending on the region of the chromosomes disrupted, include hearing, vision, cardiac, renal, neurological, urogenital, skeletal and endocrine disorders as well as major and minor birth defects.

What are the challenges our families face?

The main challenges are:

1. Rarity
2. Negative family feelings
3. Difficulties in describing the child's disorder
4. A complex set of symptoms to deal with
5. A lack of information
6. The large number of professionals involved
7. Stress on the family

1. Rarity

The chromosome disorders with which we deal are individually rare, sometimes unique, but collectively numerous. About 1 in 200 babies is born with a rare chromosome abnormality. Many of these will have what are know as 'balanced' chromosome rearrangements and will find out about them only when they have problems having their own babies in adulthood: fertility problems, multiple miscarriages, stillbirth or the birth of a disabled child. About 1 in every 1000 babies will be born sick and disabled because of an unbalanced chromosome abnormality.

2. Negative family feelings

On being given a diagnosis, families may experience feelings of isolation, bewilderment, shock, desperation, guilt, loneliness, physical and mental stress and bereavement – both literal as well as feelings of bereavement for the 'normal' child they expected to have.

> *After we were told about our son's condition, we were told to take him home and just love him. There was no help for me and I just wanted a way out – the first couple of years were indescribable. I was in so much pain inside and there was nobody to talk to who would understand.*
>
> A parent.

3. Difficulties in describing their child's disorder

Most chromosome disorders are so rare that they have no 'syndrome' name but are described by an abstruse code consisting of a series of letters and numbers. How do professionals explain these results to families? How do families describe their child's chromosome abnormality to other people? Imagine trying to explain to family and friends what your baby's disorder is when the only name for the disorder is a complex technical code (called a karyotype code) like this!

> **46,XY,t(3;7;12)(3pter->3p12.2::7q31.3::3q25->3p12.2::7q21.2->7q31.3::3q26.3->3qter;7pter->7q21.2::12p12.3->12pter;7qter->7q32::3q25->3q26.2::12p12.3->12qter)**

This challenge is likely to become even worse as more sophisticated types of molecular analysis, detecting much smaller changes in an affected person's DNA, become mainstream.

4. A complex set of symptoms to deal with

As already alluded to, with a unique and wide range of genes coding for different body systems being deleted, duplicated or rearranged with each individual rare chromosome abnormality, the person affected by the disorder will have a complex set of symptoms. These might include various combinations and permutations of learning and physical disabilities, developmental delay, medical issues, dysmorphic features, challenging behaviours and/or mental health problems. Many people affected by a rare chromosome disorder will develop late onset conditions as they enter adolescence and adulthood.

5. A lack of information

For most rare chromosome disorders, there has frequently been little or no information to be had. Even genetics professionals may not be able to provide specific information and will often only be able to give families generalities and possibilities about their child's prognosis, often with worst case scenarios or the recommendation to 'wait and see'. This is incredibly depressing and worrying for affected families and frustrating for the multitude of professionals who might be involved in the child's case. They often have no guidelines as to how the child might be affected, what conditions to watch out or monitor for or what therapies and care management pathways would help the child reach their full potential.

> *Outlook provided a very bleak picture indeed. Answers even to basic questions on lifespan were vague with no promises or hope on anything. I left this meeting with the paediatrician feeling full of despair for the future.*
>
> A parent

6. The large number of professionals involved

With the complex nature of the symptoms arising with rare chromosome disorders, another big challenge for many affected families is the large number of professionals involved in their child's care and in the family's welfare. It is not uncommon for an individual family to see hundreds of different professionals over the course of their child's lifetime. Excellent communication and keyworking are paramount if families are to cope with caring for their very special children, the Team Around the Child approach being ideal for many families. Sadly, many families have to contend with poor communication between themselves and professionals and many do not have an assigned keyworker to ease the burden. Many mums and dads find themselves acting as their child's keyworker.

7. Stress on the family

Without the right support, many families break up under the pressure of living with a child with a rare chromosome disorder. Some parents develop problems with their mental health and well-being. They very often lose friendships and other relationships and it is not uncommon for them to experience financial and employment difficulties, especially if their child needs to go into hospital a lot or to attend a multitude of different appointments on different days. There is no doubt for many families, rare chromosome disorders bring about a significant lifestyle change. Then there is stress on other family members – the affected child's siblings and their grandparents can suffer too.

> *I have become quite ill and depressed by having to carry the guilt of my balanced translocation around. I wish my son and I could have died the day he was born.*
>
> A parent.

It doesn't take much imagination to realise that co-operation between professionals and parents is essential to meet these extraordinary challenges and to help families cope with caring for their unique child.

Jenny Searle's story

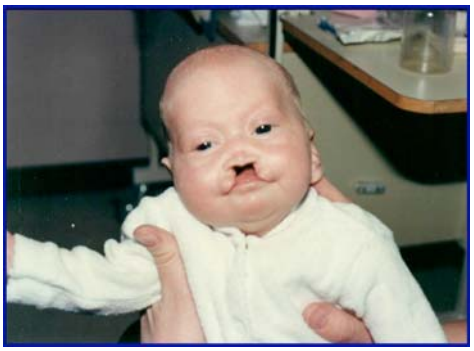
Despite all her very complex special needs, Jenny is a delightful, sociable young lady who loves having her friends round her, going out to exciting places, as from May this year living in her own home with friends (albeit with 24-hour nursing care), listening to music and 'gossip', having her make-up and nails done…Indeed, socially she's very much a typical teenager!

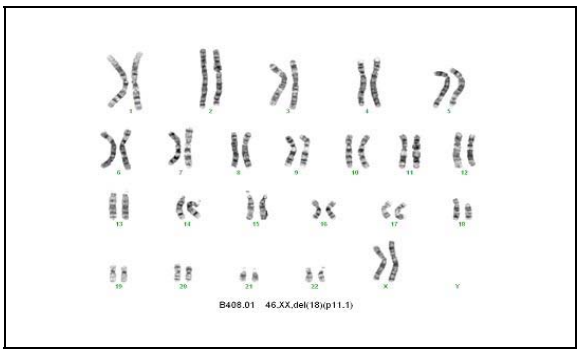
Jenny

To illustrate how complex some children with a rare chromosome disorder can be, I would like to tell you more about my daughter Jenny. She has lost the short arm (p) of chromosome 18. (See the photo of Jenny's chromosomes below and note one copy of chromosome 18 is reduced in size.) Jenny was born at 33 weeks gestation and her arrival, after the birth of my first healthy, bright child, was a great shock. As you can see from the photo of her as a baby below, she had a severe midline facial cleft lip and palate and was (and still is) missing the bone structure to her nose.

Jenny has very complex special needs, being profoundly physically and intellectually disabled. She has no formal communication skills and no self-help skills whatsoever. She is totally dependent on her carers and on technological aids, including a moulded wheelchair, hoists, suction machines, nebulisers, a profiling bed, gastrostomy feed pump, incontinence aids, ileostomy bag and van with a wheelchair lift. She has complex congenital and acquired health issues including epilepsy, hypopituitarism, hypothalamic dysfunction, holoprosencephaly, midline cleft lip and palate, gastro-oesophageal reflux, chronic respiratory problems, otitis media, scoliosis, bilateral hip dislocations, multiple fractures due to acquired osteoporosis, malignant lesions in her colon, etc. etc. She has had numerous surgeries, medications and therapies.

When Jenny was born, there was no family-friendly information available about her rare chromosome disorder, only a few stark, frightening and very negative articles in the medical literature. I was very relieved though to be put in touch with Edna Knight and her fledgling rare chromosome disorder support group. It was such a comfort to know we were not alone.





Jenny at birth and her chromosomes

As a family, we could have gone downhill fast after Jenny's arrival but we have had fantastic support over the years from a large, dedicated multi-disciplinary team of professionals with excellent keyworkers as well as the superb support and information provided by *Unique* – it has made all the difference. Thanks to all these wonderful people, we have managed to maintain as normal a family life as possible, both parents being able to work, Jenny's brother turning out to be a well-balanced, intelligent young man now studying medicine and, most importantly of all, Jenny being happy, content and much loved and liked in her community. If a family like ours with such an extremely disabled and sick child can be helped to stay together and flourish with the right support, then why should this not be possible for all families affected by a rare chromosome disorder?

How can *Unique* help?

Unique has a staff team of seven, two full-time and five part-time, and well over 150 volunteers. All our staff members are parents of children with a rare chromosome abnormality; some of our children are severely or profoundly disabled; sadly two of us have lost our children. We offer our professional and personal expertise, knowledge and connections to assist our members. By great coincidence, two of us were post-doctoral research geneticists before having our children while another is a highly-regarded medical journalist. Between us we speak several languages fluently besides English: French, German, Punjabi, Hindi and Kiswahili, as well as understanding some Bulgarian, Urdu, Pushtu and Gujerati.

We run telephone and email helplines for families and professionals to pose their questions about rare chromosome disorders or any related issues. We try to soften the impact of a diagnosis of a rare chromosome disorder by offering links with affected families, providing reliable, validated information and by raising public awareness. Over ten years we have developed a highly innovative database of rare chromosome disorders with karyotypes or microarray results together with medical, social, developmental, behavioural and educational information on thousands of affected individuals. The database is constantly updated and anonymised information used to inform families and professional members, providing a cost-efficient resource that makes a huge difference to a large number of people. The database has allowed us to observe and record the natural history of many chromosome disorders and to counterbalance negative presumptions with families' positive experiences.

We have produced more than 100 pioneering family-friendly, medically-verified information leaflets on specific rare chromosome disorders. Many more leaflets are in the pipeline and some have been, or are being, translated into other languages by colleagues in Europe and the USA. The depth and extent of our information is not available anywhere else. We have developed a comprehensive website (www.rarechromo.org) from which our information leaflets can be downloaded freely.

We have an active presence on social networking sites such as Facebook and MySpace and publish a members' magazine of high quality three times a year. We hold regular conferences at which families and professionals can meet and exchange information and ideas, and get to know and understand each other in a social setting. We run study days on specific chromosome disorders where families and experts in the disorders can meet and establish common themes to contribute to the knowledge about the disorders and to stimulate research into these disorders. Building on our collective knowledge, it is our intention to formulate care management pathways for each specific disorder to enhance the life chances of each affected member.

Note: If you need explanation of any of the medical terms used in this article please contact Beverly or IQJ's Editor.

How to contact us and learn more: If you would like to learn more about rare chromosome disorders and their effects, please view the information on our website. If you are affected by a rare chromosome disorder and would like to join *Unique* or if you are a professional with a patient or client affected by a rare chromosome disorder, please feel free to contact me at:

Beverly Searle PhD, Unique CEO
PO Box 2189, Caterham, Surrey, CR3 5GN
Tel: 01883 330766

E-mail: info@rarechromo.org Website:
www.rarechromo.org