THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

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N E W S L E T T E R

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MISSION STATEMENT

To facilitate support for those affected directly or indirectly by genetic conditions throughout Australasia.

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EDITORIAL

No one will forget the insane acts of 11th September 2001 that changed the world forever. The impact has been immense. Our thoughts go to the families and individuals who have lost loved ones and we hope for worldwide visionary leadership in a time of crisis.

AGSA has had a busy couple of months running seminars.

AGSA's AGM on 21st October 2001 saw the election of our new committee. I would like to welcome our new committee members, Scott Brightwell, Melanie Cameron - Lismore,Ann Mulder – Lismore, Kim Palmer, Roberta Perla, Richard Petrie, Gayathri Parasivam, Carolyn Shalhoub, Lilea Sward, Karen Schuler and Kris Barlow- Stewart as Public Officer. We would like to thank our guest speaker Dr Kris Barlow-Stewart for her thought provoking talk and discussion on privacy issues.

Twenty people attended AGSA's Triple X syndrome seminar on 14th October resulting in a future family social get together, an annual meeting and a newsletter. It is interesting to note there are possibly 9000 women with Triple X Australia wide and 3000 of these are in NSW. During the day an issue regarding prenatal testing

was tabled. Sex chromosomal anomalies are not mentioned as a possible outcome of prenatal testing and when the result was positive for one of these conditions termination was the first issue discussed. The abruptness and timing was felt to be inappropriate and insensitive All present agreed the risk of detecting one of these conditions should be mentioned by the doctors and genetic counsellors before the test is given. It was an excellent day with many issues covered and friendships formed.

The Breast Cancer Genetic Testing Information Seminar on 7th October was attended by twenty-two people and was a very rewarding day. This was a first meeting of its kind. All present agreed to hold an annual meeting in October at AGSA. It was agreed to not set up a formal support group as they felt they need the input from professionals because of the rapid changes in genetics and the emotional impact of cancer.

For 2002 AGSA is hoping to be more proactive on genetic issues such as discrimination, privacy, insurance etc. with the assistance of our Medical Advisory Board.

Please send me information about your group if you wish to put it on AGSA's up-dated web site **www.agsa-geneticsupport.org.au**. Thank you to those groups who have sent me details of conferences and links. We are happy to support your group through our web site.

Until next issue
Best wishes
Dianne Petrie

"Making the right connections since 1988"

AGSA would like to apologize for the lateness of this newsletter due to circumstances beyond our control.

Parents spend the first part of a child's life getting him to walk and talk, and the rest of his childhood getting him to sit down and shut up. Anonymous

Officials from the Association for Genetic Support of Australasia (AGSA) met with Jannine Cody, President of Genetic Alliance, in Sydney. From left to right: Richard Petrie, Dianne Petrie -AGSA Peer Support & Information Officer, Jannine Cody and Scott Brightwell.

LETTERS TO THE EDITOR

Thank you for hosting the Rare Chromosomes Disorder Seminar last Sunday. I had always thought of Cornelia de Lange Syndrome as being rare, but I discovered its really quite common in comparison to the unique disorders represented at this meeting.

When my son was diagnosed 18 years ago, I was told there weren't any others like him, and to not bother looking. How glad I am that I ignored that advice. Being in touch with other families with like problems has been THE most helpful tool we have had in raising our special child. I can only imagine how difficult it is for those families who literally have no one else to talk to who truly understands. If it weren't for umbrella support groups like AGSA and Unique Australia, these parents would surely be alone.

It was interesting to discover the common medical problems our children face, despite their different genetic conditions, and how much the parents appreciated the support and understanding of the

medical specialists who had helped them over the years.

I appreciate being included as a delegate at this seminar, and would like to encourage more parents to take advantage of attending similar functions, even though their child's syndrome doesn't exactly match the host group. I have gained further understanding of other conditions, genetics and health care, which is of use to both my son, and to the CdLS families who I represented. Not to mention the wonderful people I have had a chance to meet!

Thanks especially to the doctors who gave up their Sunday, and time with their own families, to help others.
Jenny Rollo
CdLS Association Inc

AGSA Committee Members for 2001-2002

Carolyn Shalhoub
Carolyn is very excited to be joining the AGSA committee. She is an Associate Genetic Counsellor at Royal North Shore Hospital and has been working there for about three and a half years. Prior to becoming a genetic counsellor, Carolyn spent many years working on the other side i.e. in a laboratory doing genetic research. She currently works very closely with the antenatal clinic at the hospital and thus most of her counselling relates to prenatal diagnosis.

She loves her job and is very passionate about genetics and its impact on people. She also feels very strongly that all individuals, couples and families are adequately informed in order to make choices that are right for them, and that support is paramount throughout the entire process.

Karen Schuler
Prior to having her three children, Karen worked in the finance industry while studying bookkeeping and accounting. After the birth of her second daughter, Karen became a stay at home mother. Once her two daughters were at school, she undertook a major career change and studied for four years to become an Occupational Therapist.

The birth of her son, Alexander with mosaic Trisomy 18 in 1994 triggered Karen's real career change. She currently juggles caring for Alex, being a grandmother and working with others. She co-ordinates SOFT Australia, a support group for rare Trisomic disorders, which she founded in 1996. She is a board member for a number of groups who advocate for children with special needs and also works part time as a medical researcher.

Recreationally Karen enjoys gardening, walking and despite being scared of computers, web designing.

Kim Palmer
Kim currently works as a freelance journalist while looking after her two sons. Oscar was born in 1998 with the rare disorder of a partial duplication on chromosome 4. His brother, Felix, followed in 2000. While Kim and her husband Andrew, were fully aware of all Odd Couple references, the boys are very different to each other.

She served on the AGSA committee in 1999 and 2000 and her passion for social equity, raising community awareness, interest and knowledge on genetics and how the never-ending discoveries about our genome will impact all our lives has drawn her back into the AGSA fold.

She loves to cook and will very rarely pass up an offer for a drink, feed or a chat. She dreams about nights of uninterrupted sleep, days with no form of speech, physio or occupational therapy and an income solely dedicated to eating out, shoes and overseas glossy magazines.

Lilea Sward
Support Group/Condition represented: CAH - Congenital Adrenal Hyperplasia. *(see details of CAH at the end of the committee profiles).* Lilea has knowingly lived with a genetic condition since she was five years old. Her condition is recessive (both her parents carried a DNA mutation and showed no symptoms). She has an older sibling also affected. Lilea lives a very normal everyday life to a large degree but takes medication daily to provide the hormones her

body does not produce due to an enzyme deficiency (21 Hydroxolase - affecting approximately 80% of CAH cases). Since belonging to a Support Group and AGSA, the ability to meet others with similar or the same condition, to interact with other individuals who have dreams - just like her, hassles in life- just like her and goals, somewhat like hers has changed her life dramatically - for the good. She believes life is not a spectator sport so.....Dream it....Plan it....Live it!

Website - CAHSGA (CAH Support Group of Australia)
http://home.vicnet.net.au/~cahsga/

Gayathri Parasivam
Gayathri have been working in molecular research prior to completing Part I of Genetic Counselling training. Her experience therefore was lab based, and although she enjoyed the science, she was interested in taking this into the clinical realm and having the patient contact. This has led her to genetic counselling. Gayathri is currently employed in Clinical Genetics at Royal Prince Alfred Hospital, and she is finding the role both challenging and rewarding. The variety and sometimes unpredictability of patient responses is both interesting and enjoyable.

The change of job has meant a move to Sydney from Melbourne. So far she is finding that the upheaval has been well worth it, and is looking forward to grounding her feet for a while. She is certainly looking forward to broadening her experience in genetic counselling.

Scott Brightwell
Since 1972 Scott has worked in the Customs Brokerage, Freight Forwarding, Commodity Trading & Logistics areas. Included in this time was six years on the Senior Executive Committee of a publicly listed company. He currently holds the position of State Manager NSW, of a medium sized Australian owned Customs Brokerage / Freight Forwarding company employing in excess of seventy staff.

He and his wife Marlene have two daughters Allison (18) and Erin (17). Erin has 18q deletion syndrome.

Scott served on the AGSA committee 2000-2001, and was a foundation member of the Rare Chromosome Disorders Group. He transferred to The Chromosome 18 Registry and Research Society of Australasia when it was formed in 1997.

Richard Petrie
Richard has been on the AGSA committee for about eight years and believes the organization has a unique role in meeting the needs of a significant segment of the community.

Richard has a daughter with Williams syndrome, now aged eighteen and at the time of diagnosis doctors knew of only two other families in Australia with a WS child. There was little information on the condition and very poor information on what services were available. Everything seemed very difficult. With his wife, Dianne, they set up the Williams Syndrome Support Group in 1985, which now has over 200 members nationally. Richard believes if AGSA had been in existence many aspects of living with a child with a disability could have been so much easier.

Richard has a background in pharmaceutical sales and marketing and works for Bayer Pharmaceuticals.

Melanie Cameron- Regional Contact

Melanie is the mother of seven children. Sadly, her first son, Drew, died at 17 months in April 1999 of an unknown type of mitochondrial disease. Since then she has established the Aussie Mito, United Mitochondrial Disease Foundation Australian Chapter and is in contact with many families.

At present she is busy organizing a Musical in the Park.

Melanie believes in the value of contact with others and the valuable role of support groups. She is looking forward to organizing an outreach seminar with AGSA in 2002.

Roberta Perla

Roberta is a clinical psychologist who has had extensive experience in the community health setting for the past 10 years.

Her special interests are in research into ADHD and the physiology of brain development as well as working with adolescents and adults with educational and substance abuse problems.

Ann Mulder - Regional Contact

Ann is married to Chris, a civil engineer and they have five children, four boys and one girl, ranging in age from six years to fifteen years. Two of the boys have the genetic syndrome Pseudohypoparathyroidism; also known as Albrights Hereditary Osteodystrophy.

Ann grew up in Sydney and Lismore, in northern NSW, before moving back to Sydney to study pathology at university. She worked as a scientific officer at the Prince Henry Hospital,Randwick, mainly in Biochemistry and Haematology.

Ann and Chris travelled to Asia and Europe for two and half years before starting their family.

After returning to Lismore, Australia whereDavid their eldest,who is now 15 years old, was born. Shortly after, 13 months to be exact, they had Mark who has Pseudohypoparathyroidism plus aspects of Aspergers syndrome. Three years later, Luke was born. He also has Pseudohypoparathyroidism - later Nicholas and Gabrielle were born. When Luke was a baby they moved to Fiji on an Aid Project staying for five and half years. It was a wonderful experience.

When Mark and Luke were nine and six years old they were diagnosed with their syndrome. Shortly afterwards they moved back to Australia in search of better medical care and hopefully more support.

During the next four years they started a support group for Pseudohypoparathyroidism, and have spent many hours researching their condition.

Ann is happy to say the boys' potential now exceeds all expectations. They attend a mainstream school, can read and continue to learn well. Mark is an exceptional guitarist and it looks like Luke will follow in a similar fashion. They are both beautiful boys and continue to astound and teach their parents, with their persistence and hard work. This year their youngest, Gabrielle started school and Ann has gone back to University in Lismore to study full-time for a Bachelor of Naturopathy. She has enjoyed the stimulation of studying enormously. Her future as a naturopath is to hopefully work with people with learning difficulties, autism, and cognitive impairment as well as research into the role of herbs and brain function.

Ann is looking forward to being a part of the committee of AGSA and she hopes to contribute in a constructive and helpful way.

CONTACT CORNER

AGSA will publish requests for contacts and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write directly to the individual or group concerned if an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-

judgmental approach is recommended in establishing contact.

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Ataxia Telangiectasia

A lady would like contact with others with this condition. Please contact AGSA for details.

****

Duane syndrome

A rural family is looking for contact with others.

****

46XY del (10) (q23.2q24.1)

A mother of a three-year-old boy would like information and contact with a family with a child with a similar condition.

****

Kearns-sayre syndrome

A mother of an 11-year-old boy with this condition would like contact with another family.

****

Klippel-Trenauney syndrome

AGSA has two families with this condition who are looking for contact with others.

****

Lowe syndrome support group – contact AGSA if you wish to be involved in this group.

****

Metachondromatosis

A Victorian lady would like contact with others. Contact AGSA for details.

****

Mitochondrial Disorders

Two families, **Cytochrome C Oxidase Complex IV deficiency** and **Kearns-sayre syndrome** would like contact with others. Please contact AGSA for details.

****

Hyperprostaglandin E syndrome

A Queensland family would like contact with others.

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X-linked Hypophosphatemia

A mother would like to organize a get together next year with other families who have a child with this condition. Presently she knows of five families. If you know of any families who may be interested please ask them to contact AGSA for further details.

***Most of you know of or have heard of, Dr Chris Green, Paediatrican**, the author of **Toddler Taming** and how he is subsequently, a special person to many parents. It is therefore saddening to hear he had suffered a stroke after heart surgery, which left him unable to speak. Those who heard him speak at the inaugural Genetic Disorders Awareness Week in 1997 will know what a cruel blow this is. I met Chris when he agreed to talk at the first National Williams syndrome conference at North Head, in 1990, and again, at the above mentioned launch. As Chris has supported and given so much to so many people including myself, I decided to write something here so that others, touched, enthused and encouraged by his words, may be able to send their wishes and encouragement to him. Most of us will always remember the many many times his advice and insights into children made us laugh, and should take comfort when he writes "life has not turned out as expected but I am well and busy".*

If you would like to send a message to Chris, please forward it to AGSA and I will pass it on. Thank you. Dianne Petrie

Bloody Doctors!

Christopher had had time off work to finish writing "Beyond Toddlerdom." Before returning to work, a routine medical checkup discovered that the covering of his heart was constricting it. It seemed a good idea to get the problem fixed! All went well until after the operation when a clot

formed in the heart. Some of it broke away and caused a stroke.

On the evening before the operation, Chris had just completed the book. Now, within 24 hours of the stroke, he was left with the ability to say only two words. It seems really cruel that the language center – the part of the brain that controls speech, reading and writing….was the main part that was affected. The very skills Chris was known for were taken from him.

Effects of the Stroke

There has been good recovery but since Chris's work involved communicating, and his brain is still full of ideas and knowledge, it is very frustrating that he can no longer communicate. In his own words, he can say, "Bugger nothing!"

Working

Of course, Chris would still love to be working at the Children's Hospital, Westmead, with his team, who had all worked together for so long. But it was becoming obvious that this was not going to happen. In June 2000 he officially retired from the Hospital.

Retirement

Despite this he has not been sitting around idle. He has spent the last six months revising his books "Babies" and "Toddler Taming".

Working on the books has been difficult – about 20 times harder than it used to be. Chris has always wanted the words to be absolutely right and as he has to work with someone to get his words onto paper, usually his wife - it is even harder to get them out and it causes a lot of stress.

For six months after his stroke he couldn't see any purpose in living. Being busy had always been important to Chris and he coped by continuing in this way. He has painted the house inside and out and has become particularly good at washing and ironing! Whenever words are too difficult he runs away……. Chris loves solo sailing and cycling, staying in hostels as he goes. For instance, last year he cycled around Tasmania and walked the mountain tracks of New Zealand.

Message for Parents

"As somebody who so nearly died, I wonder what people would remember me for?

When I reflect…I didn't split the atom or discover the cure for cancer, but I think I helped a lot of parents. Many parents whose children were difficult sleepers found that within three days my techniques had changed their lives. I hope parents realize that their toddler is not the enemy, to take time to enjoy him and to see the fun and magic in their child.

Life has not turned out as expected but I am well and busy".

Chris.

Any messages for Chris contact AGSA.

PROFILE
A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes that where overlaps occur in conditions, support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.

Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is "Alive and Well' and happy to take referrals.

TRIPLE X SYNDROME

Also known as:

- 47,XXX Syndrome
- 47,XXX Karyotype
- 47,XXX Chromosome Constitution
- Trisomy X

General information about Triple X syndrome:

Triple X Syndrome is a chromosomal problem that only affects females. Females normally have two X chromosomes; however, those with Triple X Syndrome carry three X chromosomes (*trisomy X*) in the nuclei of body cells. No specific pattern of symptoms and physical features (*phenotype*) has been found to be associated with this chromosomal make-up (ie. 47,XXX karyotype).

Many affected females appear to have no or very few associated symptoms, while others may have various abnormalities.

However, investigators indicate that Triple X Syndrome is a relatively common cause of learning difficulties, particularly language-based disabilities (eg. dyslexia), in females. Evidence suggests that affected females typically have normal intelligence with IQs that tend to be lower than that of their brothers and sisters (*siblings*). Mental retardation rarely occurs. Infants and children with Triple X Syndrome tend to have delayed acquisition of certain motor skills and delayed language and speech development.

Affected females often are of tall stature. According to researchers, although sexual development and fertility are usually normal, some may have delayed puberty and/or fertility problems. In addition, in some cases, certain physical abnormalities have been reported, such as a relatively small head, vertical skin folds that may cover the eyes' inner corners (*epicanthal folds*), and/or other findings. Triple X Syndrome results from errors during the division of reproductive cells in one of the parents.

The syndrome results from errors during the division of reproductive cells in one of the parents.

Symptoms of Triple X syndrome in more detail:

As noted above, researchers indicate that no specific pattern of symptoms and malformations (*phenotype*) appears to be characteristically associated with Triple X Syndrome. However, Triple X Syndrome has been determined to be a relatively common cause of developmental and learning disabilities in females.

Reports indicate that some affected females may tend to have a relatively low birth weight that is within normal limits. However, many develop an above average height during childhood as compared with others their age, tending toward tall stature in adulthood (e.g., above the 75th percentile).

In contrast, in some affected females, the head circumference may tend to be relatively smaller than expected for their age and gender. In some instances, additional physical findings have been reported, such as:

- unusual shortness and broadness of the head (*brachycephaly*);
- vertical skin folds that may cover the eyes' inner corners (*epicanthal folds*);
- widely spaced eyes (*ocular hypertelorism*);
- abnormal deviation of one or more fingers or toes (*clinodactyly*);
- widely spaced nipples;
- other findings.

In most cases, sexual development and fertility are normal. However, reports indicate that some affected females may have abnormal development of the ovaries (*ovarian dysgenesis*) and/or the uterus; delayed puberty; and/or fertility problems. In addition, kidney abnormalities, such as absence of a kidney (*unilateral renal agenesis*); recurrent urinary tract infections; and/or other abnormalities have also been reported.

As noted above, evidence suggests that females with Triple X syndrome typically have normal intelligence with IQs that tend to be lower than that of their siblings (e.g., approximately 10 to 15 points lower). Mental retardation rarely occurs.

(To be classified as having mental retardation, an individual must have an IQ that falls below 70.)

As early as infancy, mild developmental delays and learning difficulties may be apparent. For example, affected infants and children may have decreased muscle tone, poor coordination, awkwardness, and delayed acquisition of certain motor skills. In addition, delayed language and speech development (eg. delays in receptive and expressive language) may become apparent by approximately 12 to 18 months. Reports indicate that females with Triple X syndrome have an increased frequency of reading disorders, including dyslexia or other language-based learning disabilities.

In some cases, only a certain percentage of an affected girl's cells may have three X chromosomes, while others have a normal chromosomal make-up (46,XX/47,XXX mosaicism). Evidence suggests that such cases are associated with mild symptoms and fewer developmental and learning problems. Variants have also been described in which cells contain four or five X chromosomes (Tetra X Syndrome and Penta X Syndrome). Such variants are typically associated with more severe symptoms and findings.

What causes Triple X syndrome?:

Triple X syndrome is a chromosomal abnormality in which there is an extra X chromosome. Chromosomes are found in the nucleus of all body cells. They carry the genetic characteristics of each individual. Pairs of human chromosomes are numbered from 1 through to 22, with a 23rd pair that normally consists of an X and Y chromosome for males and two X chromosomes for females. Thus, females with a normal chromosomal make-up (*karyotype*) have 46 chromosomes, including two X chromosomes (46,XX karyotype); they receive one chromosome from the mother and one from the father in each of the 23 pairs.

However, females with Triple X Syndrome have 47 chromosomes, three of which are X chromosomes (47,XXX karyotype). The presence of the extra X chromosome results from errors during the division of reproductive cells in one of the parents (nondisjunction during meiosis). It is believed that the extra X chromosome is received from the mother in most cases.

Who is affected by Triple X syndrome?:

Triple X syndrome is a chromosomal disorder that affects only females. Reported estimates concerning the disorder's frequency have varied, ranging from one in 1,000 female births to approximately one to two in 3,000 female births. However, because many females with the disorder may have few or no symptoms, they may never be diagnosed with the disorder. Many researchers suggest that the disorder is in fact under diagnosed and that the reported number of cases as reflected in the medical literature is inappropriately low. Due to such factors, it is difficult to determine the true frequency of Triple X syndrome in the general population.

Is there any treatment for Triple X syndrome?:

Diagnosis

Triple X Syndrome is diagnosed based upon chromosomal analysis that reveals the presence of an extra X chromosome in body cells. It is usually detected unexpectedly in females with suspected developmental and learning disabilities. In addition, Triple X Syndrome is increasingly being diagnosed before birth (*prenatally*) based on chromosomal analysis performed using amniocentesis or chorionic villus sampling (CVS). During amniocentesis, a sample of fluid that surrounds the developing fetus is removed and analysed, while CVS involves the removal of a small sample of the placenta.

Treatment

Early intervention services are recommended for infants and children diagnosed with Triple X Syndrome. Experts advise developmental assessment by age six months to evaluate muscle tone and strength; language and speech assessment by age two years to evaluate expressive and receptive language development; and reading assessment by school age to rule out or confirm dyslexia. Evidence suggests that

affected children are greatly responsive to early intervention services and treatment.

Because females with Triple X Syndrome have a slightly increased risk for chromosomal abnormalities during pregnancy, prenatal counselling is recommended.

Rare Chromosome Support Group
C/- Association of Genetic Support of Australasia (AGSA)
66 Albion Street
SURRY HILLS NSW 2010
Ph: (02) 9211 1462
Fax: (02) 9211 8077
Email: agsa@ozemail.com.au
Home Page: http://www.agsa-geneticsupport.com.au

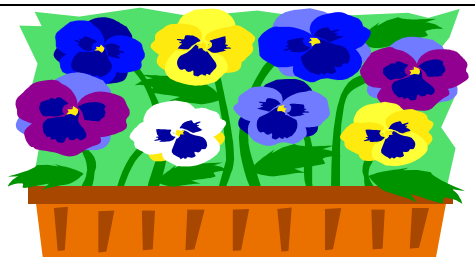
The following overseas groups may be able to provide additional information and support:

Rare Chromosome Disorder Support Group
160 Locket Road
Harrow HA3 7NZ UK
Tel: 0181 863 3557

References:

This information sheet is based on information available from NORD (see below) which was current in October, 2001.

NORD
(National Organisation for Rare Disorders)
PO Box 8923
New Fairfield, CT 06812-8923
Telephone: (203) 746-6518
Fax: (203) 746-6481
Email: orphan@nord-rdb.com
Home Page: http://www.nord-rdb.com/~orphan



FAMILY STORIES

The following personal stories were presented at AGSA's Triple X syndrome Seminar and Breast Cancer Genetic Testing Seminar held recently on consecutive Sundays. I feel privileged to have been a part of such generosity of spirit. Dianne Petrie.

Triple X syndrome

Dean and Janice 'A parent's perspective'

We have two children Ben aged nine and Sophie aged one year. We know little about Triple X but would like to share our experiences about our two children.

Ten years ago we decided it was time for a family and within 6 months I was pregnant and looking forward to having a baby. When I was 24 weeks pregnant I went into labour. I was taken to Wagga Base Hospital and told I would have to be flown to Sydney as Wagga didn't have the facilities for a baby born under 30 weeks gestation.

We were told that there would be a big chance that our baby would not survive but if he did survive, they showed us pictures of what he would look like and all the machines that would be attached to him to keep him alive.

I delivered Benjamin Charles McDonald naturally, 15 weeks early on 12/12/91, (his due date was suppose to be 23/3/92); he weighed 850 grams or 1 lb 14oz. The Doctor at one stage told

Dean to get me to have a look at our baby, as he wasn't expected to live. They gave Ben a drug called surfactant that lubricated his tiny little lungs. This did not work and they gave him more of this drug and his lungs responded.

He was in intensive care for 101 days at the Royal Women's Hospital at Paddington. During this time he was transferred to another hospital where he had surgery to repair a hernia. He had lots of set backs due to infections, 5 blood transfusions and other numerous complaints. He was transferred back to Wagga Base Hospital and after 108 days in hospital we finally got to bring our beautiful little boy home. He weighed 4lb 11oz at this time. Today he is a very normal boy with no disabilities at all. He is very coordinated and is extremely good at any sport he plays.

Ben was quite a handful for some time and it wasn't until three years down the track that we decided to have another baby. I had five miscarries in approx. five years, the last being twins. We enrolled on the IVF programme in these five years to speed things up as I was getting older and falling pregnant was getting harder. After only one pregnancy through IVF, we decided that we would try something else.

We found out about a naturopath in the New Idea magazine and her success with infertile women. One story in particular that we were interested in was a woman that had five miscarriages. After seeing her there was a successful pregnancy.

This was going to be our last shot at trying for a baby. She looked into both our eyes and noted problems and gave us herbal mixtures to take as well as an array of herbal tablets. We went on a very strict diet to detoxify our bodies, this diet took 12 weeks; we then went back up to see her in which we started on a fertility programme, which was in the line of herbal mixtures and tablets. Within nine months I was pregnant. She then sent me down more mixtures and tablets to help me carry the pregnancy. After twelve weeks had come and gone we were ecstatic that I had got passed this danger stage. We finally had felt confident that every thing would be OK.

At fifteen weeks I went to Canberra and had an amniocentesis, as I was now 38. At seventeen weeks my Doctor rang me to say the results had come back and they were abnormal. I was devastated; I couldn't believe that this could be happening. He told me that we would have to see a Genetic Counsellor to explain our options, one being termination. We had to wait three days to see this Genetic Counsellor and it was the hardest three days of our lives.

She explained to us that our little girl had an extra chromosome and it was called Triple X syndrome, she went on to say that she would live a normal life, the baby would look like us, would have children of her own but would have difficulty with maths and some other subjects at school. We walked out of there convinced that this was definitely not bad enough to terminate her and would deal with these problems as they arose.

I rang our naturopath as soon as we got home from Canberra and she sent me down herbal mixtures and tablets that I had to take that would be good for the baby's brain, she also told us to name our baby and talk to her all the time whilst I was pregnant. We had to go back to our Doctor one week after seeing the Genetic Counsellor with our decision. When we told him that we were keeping the baby he asked us twice 'are you sure that is what you want to do'.

I left there feeling very down and unsure of everything. All I wanted to do was to talk to some parents of these children for their views. I was given Dianne Petrie's phone number and rang. I broke down crying to her. She was fantastic and gave me other families' phone numbers. Both Dean and I rang and listen to what each had to say. They put our minds at rest. We thank you both for your advice, which helped us cope with the coming months ahead.

I must admit, this Triple X played on my mind the whole time I was pregnant and I really didn't enjoy my pregnancy. With Ben born so early I didn't feel as though it was a pregnancy, I wasn't even showing at twenty-five weeks. I busied myself with work so that I didn't have to think about it. I worked up to three weeks before my

due date but she decided to come two days after I finished work. I was booked into a Canberra hospital to have her but as this was two hours away, the doctor in Tumut decided on Wagga Hospital. We got there at 4.30 am and Sophie Jo McDonald was born at 7.00 am weighing in at 6lb 2 oz. Our son, Ben was present as well as my mother, Sherrin, for the birth. I was very happy to think that I went full term and had the chance to have my baby given to me after she was born, as this was not the case when I had Ben, but I still worried about the condition she had.

To-date she has met all her milestones. She is now walking. She is a very quiet baby, never crying unless she is tired or wants a bottle. When she wakes up she plays quietly in her cot, which concerns me a little. We all love her so much and she has given us so much joy, and we will deal with her problems if they arise.

In one way I wish we never found out about Sophie's condition as I do worry about it, but on the other hand we have been given a chance to address the problems as they arise and try to help her stay ahead.

Thank you for listening to our story.
Dean and Janice

SALLY'S STORY

BREAST CANCER

I had breast cancer diagnosed nearly five years ago aged 50, and tested positive for the BRCA 1 gene about 2 years ago. Every women for three generations on my mother's side have had breast cancer. The defect was passed down from my mother's father as both his sisters had breast cancer. This is significant for my brother's daughter as until she read a story in the tabloid press in the UK about breast cancer susceptibility being passed down the male line she had no idea that she too was at risk. My brother has since been tested and also has the defect. To my knowledge no men in my family have had prostate cancer either, though my grandfather put his head in a gas oven due to racing debts so died early and my half uncle had a heart attack. With our limited knowledge of our family history there has been no ovarian cancer.

Just to give you a bit of background to my story, my mother was diagnosed when I was 12 and away at boarding school. I was never told by my family but found out from a school friend whose mother wrote and told her "that there is so much you could do about breast cancer these days!"

Mum never hid her mastectomy scar and took great delight the following Christmas in smuggling us all watches through customs in her prosthesis! But all during her illness nothing was ever said about breast cancer, so we know nothing about any generation before her fathers. She found it very hard to cope with her breast cancer and withdrew from us all so I did not really feel I had a mother from the age of 12, until she died when I was16. I remember wondering what I was doing at her funeral and for years felt guilty that I did not miss her. I finally forgave myself when I wrote to her after my diagnosis of breast cancer. I burnt the letter and put the ashes under the bush I had planted for my sister.

The first of my sisters was diagnosed aged forty and is still going strong twenty-two years later. After this I was regularly assessed but I don't think I fully realised the significance of my family history until her twin was diagnosed aged 50. She was not so lucky and died within 18 months. At this time I contemplated bilateral mastectomies but ended up on the Tamoxifen trial instead, about which I have no regrets even though it didn't work for me.

I had no hesitation and have had no regrets about having bilateral mastectomies when diagnosed with cancer. It was only at that time doctors said that our problem must be genetic. My sister was the first to be tested. It took over 2 years to locate the defect on the BRCA 1 gene, as our defect is not in the most common position. I was devastated and cried for a week, which surprised me. But I suppose you always hope it won't be found. Obviously when I was tested the results came back quickly as they knew where to look and again I was surprised at my reaction as in my heart of hearts I knew I had to be positive as well.

Being positive for me was in some ways a non-event as I had already had cancer. My distress was for my children and what it means to them.

As a family our approach to this problem is very individual. My living sister is all for the minimalist action. She is a very strong personality and in a well-meaning way has tried to influence my decisions, but I have always acted in a way that I felt comfortable. She only had a lumpectomy (she is also singleand has decent sized breasts so would miss them) and When she found a secondary under her arm a year later, she had the lymph gland removed under local anaesthetic and demanded to be put on Tamoxifen, which worked very well for her. We've both had our ovaries removed in the last couple of years but she has had the courage or maybe the madness to go on HRT.

Our children and nieces are all coping differently. None of them wants to have genetic testing at the moment. One is on the trial of MRIs versus mammography for detection in younger women. Another has been having nipple fluid tested for a different trial. Both these nieces are heading into the danger zone as they are in their mid thirties. The latter is currently deciding whether to have another child, but later down the track is going to be tested and if positive will have bilateral mastectomies. Another niece is putting her head in the sand and hoping there will be a cure if her time comes.

My daughter is only twenty-three and very well informed. (She even had the courage to do an assignment about breast cancer for her Health Science degree). She is currently struggling through her final semester so the subject is not being raised at present though she knows I would like her to have some genetic counseling. Her main concern is about discrimination in both the workplace and insurance agencies.

I have not mentioned my son and nephews who also have to face this genetic history. Even if it does not cause them any problems, they are well aware that they, can pass on the defect, to their children. I currently have five great nieces.

To sum up, as a family we have all coped in very individual ways, which I think is important. We have to live with the consequences whatever decisions each of us make. I feel very strongly that forewarned is forearmed and even though I wish as a family we did not have this problem my sister is living proof that it can be beaten. I'm also not doing too badly myself! I am also aware that each family member has a fifty-fifty chance of testing positive, that is hard to believe this with our history of breast cancer.

EMOTIONAL ROLLER COASTER

I am very grateful to be with you all here today and to have the opportunity to share the impact that genetic testing has had on me. For me, genetic testing has been like a roller coaster of emotions. From pre-test counselling to the actual test, getting the results and the aftermath.

Due to my mum's death from ovarian cancer, I had always expected to "just have my ovaries out" when I was older. It wasn't until I arrived at the Familial Cancer Service at Westmead Hospital that I discovered we also have an extensive history of breast cancer. When my family tree was laid out in front of me, it broke my heart. It was like looking at a cemetery with rows and rows of crosses signifying my deceased relatives. Ironically that day was the first time I felt some hope I may escape the same fate.

The night before the test was agonising. I felt I was about to embark upon something that would change the course of my life regardless of the result. I didn't feel fear so much as a sense of foreboding – a sense that my life had been a series of journeys leading me toward this momentous event. Now that it was here, all I wanted to do was wrap myself in a comforting cloud of denial.

After the test, the long wait. Now that I was on the path to knowing my genetic status I just wanted the result. The idea of waiting 6 – 8

weeks seemed excruciatingly long. I oscillated between feeling sure I would be spared then pitching into depths of negativity – believing there was no way I could escape my family's faulty gene. Then there was the constant anxiety. If I was given a negative result, would I believe it? Would it not be possible that I had another faulty gene that hadn't yet been found? In that sense, a positive result would almost be a relief.

As many of you here would understand, I have been on my emotional roller coaster for some time now.

The fear of cancer isn't new to me as cancer has been a constant throughout my life, however knowing my own risk is a new fear – unfamiliar and, sometimes, over whelming. After testing positive to my families BRCA1 gene mutation and considering the options of drug trials, screening or preventative surgery, I have decided on preventative surgery as the best option for me.

I know that to some people this choice seems extreme – particularly as my risk is not then at zero. However, I sometimes feel as though I have grown up in "the shadow of cancer" After the results of my genetic test it felt like history was repeating itself - it seemed as though all the anguish my mother had suffered was being revisited on me.

My mother's death, at such a young age, has affected every aspect of my being. I have spent the whole of my adult life searching for the unconditional love I received from her, mostly in the most hopeless of places. I have jumped from one painful mistake to the next. Every fear, belief and hope I have I can attribute in some way to losing her. Now, as I near my operations, our lives cross paths again.

I remember her words and her fears written as they were on the scraps of paper she left scattered around just as I do now. My heart breaks once more. I have grieved the loss of her through the eyes of a child, young adult, and now, through the eyes of a mother. I ache realising the pain and loneliness she must have felt leaving my brother and I.

As I get older, and become more afraid of my genetic status, I find that despite fighting our similarities all these years in the hope of not following down her path, I am truly my mother's daughter. I find it very difficult not to feel guilty at the possibility of having passed on this gene to my own children – although I know that's irrational.

Like most parents, I only want to pass on wonderful things to my children – things which will enhance their lives, not potentially shorten them I resent the burden of wondering where science will be when my children are my age – what choices will they face? I mourn the possibility of them as adults testing positive and the implications that may have for their lives.

I am aware that as difficult as I have found this journey so far, the worst is yet to come. I feel like I have mourned as much as possible at this point. Although, just when I feel I've come to terms with surgery something will show me otherwise. I think I've progressed though – I no longer cry when confronted with a pair of breasts on the side of a bus advertising Bendon!

Until my mum died, my main influence was her feminist approach to life. Consequently, I've fought against seeing women as merely a set of breasts. Now though, as I face the reality of losing mine, I struggle again with how society perceives women. I wonder if I would find this easier if I were someone who would have considered a "boob job" – but I'm not.

After pregnancy and breast-feeding my breasts are stretch marked and floppy. I often joke that they are like two fried eggs sliding down my Teflon chest! But I love them and the stories they could tell! Awkward first bras, ineffectual lovers and the beautiful, warm nuzzling of my babies. I will lose the physical markers of those memories. I mourn the loss of those sensations and the realisation I will never have them again.

My hope is that through my voice and yours those around us will see us as we see ourselves – vibrant, passionate, beautiful, sexy and, above all, alive!

I'd like to finish by thanking the medical community for their ongoing dedication and research and I'd also like to acknowledge the families and friends we have loved and lost.

I am grateful for their love and wisdom. From them I hope to learn to make the presence of cancer in my life a companion rather than a guide.

The ideal committee is one with me as chairman, and two other members in bed with the flu.

CONFERENCES

IDEAS Expo 2002

24th-25th May 2002, Albury NSW

Contact 1800 029 904.

MPS National Conference

12th-14th April 2002

Sharing Our Story

Rydges Resort

Eagle Hawk Hill, Canberra ACT

For further information
National Office, MPS, PO Box
Hornsby NSW 1630
*Telephone 02 9476 841***1**

The Children's Hospital at Westmead
Education Research Institute (CHERI)

Hippocrates & Socrates V11
A powerful partnership

Disability & Diversity
Successful Living
15th & 16th March 2002

Keynote speaker
Professor Barry Carpenter, OBE,
Sunfield School, United Kingdom
Venue: John Loewenthal Auditorium

Westmead Hospital, Darcy Road, Westmead
Sydney NSW

Professional $152 for one-day attendance
$266 for two-day attendance
Parents
$105 for one-day attendance
$186 for two-day attendance
Information Contact
Leslee Edwards, CHERI, The Children's Hospital at Westmead Locked Bag 4001 Westmead NSW 2145
Telephone 02 9845 3017 Email
LesleeE@chw.edu.au

*****AGSA is looking for a Part-time Administrator, 20 hours per week. The successful applicant will have good interpersonal skills and a knowledge of the broader issues relating to disability. Tertiary qualifications in health, welfare or education and an understanding of genetic disorders are desirable. Must have excellent computer, organizational and communicative skills. Inquiries to Dianne Petrie on 02 9211 1462. Written applications to AGSA, 66 Albion Street, Surry Hills by 14th December 2001.**