# AGSA
THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

# NEWSLETTER

**October 2006 ISSUE 78** ISSN1033 - 8624 FUNDED BY THE NSW HEALTH DEPARTMENT


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**www.agsa-geneticsupport.org.au**

**MISSION STATEMENT**

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

## EDITORIAL

Thank you to all those people who attended the launch of Genetic Awareness Week on 26th July. Peter Berner was an excellent chairperson giving his time freely to support AGSA. The speakers gave am overview on Genetics: Adults Bridging the Gap and what services are available.

Lynne Brodie, Program Manger Transition Care and Dr Jane Holmes-Walker highlighted the obstacles facing patients and carers when transitioning from paediatric Care to Adult Care. A 3 – 5 year programme is currently being undertaken to identify gaps in the system and to design a model to make the transition more successful. I recently had first hand experience of this when my daughter ended up in hospital with pneumonia. It is quite different being in adult care.

Sue Pinkerton President Australian Tuberous Sclerosis Society Inc gave a brilliant talk on her personal experiences with Lizzy's care as a paediatric client to that as an adult. Sue commented it is alright for her because she is a strong advocate but not all people are like that so what do they do? Debbie Colyer spoke about her life after 18 years with PKU. This gave an insight into the complexities and difficulties of trying to have a normal life and stay on the prescribed diet. Debbie opening admitted rebelling. Thank goodness for her mum who got her on the right track again. There is an adult PKU clinic in Westmead which as been running for 9 years.

The International Genetic Alliance meeting in Brisbane as part of the 11th International Congress of Human Genetics was a great success and the talks were very interesting. The take home message was government's needs to put more into health at the beginning of life to secure a better future. The Filling the Void Project is going really well with the face to face counselling session bookings increasing.

This weekend AGSA held the 6th BRCA1/2 annual information day which was well attended.

I notice the Christmas decorations are already in the shops and of course I hope to see you at the AGM on 19th November.

Until then

Dianne Petrie

# "Filling the Void" Project.

Hi everyone - Hope this finds you all well. Can you believe we are this close to the end of the year?! Where has the time gone?

At the moment we have a group of Dad's participating in Telegroup counselling on Monday nights and a group of Mum's on Tuesday nights. These are going extremely well with lots of similarities being discovered by the members of the groups even though they are caring for children of different ages with different conditions. There has been wonderful support offered to each other through the group and has been a great source of information and advice. It is a privilege to be a part of it.

Our seminar and sibling workshop in Lismore was well attended and lots of fun. We had a social get together on the Saturday evening after the seminar for those families who wished to join us and it was lovely to share a meal, few drinks and have a chat together in an informal and fun setting.

We are in the planning stages for our seminars and sibling workshops for next year and I would love to hear from families in remote / rural areas of NSW who would like these sorts of events in your area.

We are holding a craft and coffee morning for mum's at AGSA in November which promises to be lots of fun and I will be hosting a morning tea for Multicultural Workers to try to gain better ideas for reaching families from non English speaking backgrounds.

As always, I would love to hear from any of you with ideas, suggestions, questions or just for a chat. I can be reached in the office Monday – Wednesday 9am – 5pm on (02) 9211 1462 or via email projects@agsa-geneticsupport.org.au

Bye for now

**LAURIE TAYLOR**

# CONTACT CORNER

*AGSA will publish requests for contact 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 direct to the individual or group concerned where an address is provided. The AGSA office many 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.*

**SPINK 1**
The parents of a 1 year old girl with this condition would like contact with others.

**Microcephaly Support Group**
If anyone would like more information or are trying to find a support group NSW please contact Merey Turner on 02 6682 2173.

**LEOPARD SYNDROME**
A Mum whose daughter has this condition is seeking contact with others.

# SUPPORT GROUP NEWS

**The 13th International Scientific Meeting of the Velo-Cardio-Facial Syndrome Educational Foundation, inc. in partnership with the Velo-Cardio-Facial Syndrome Foundation of Queensland, Inc is being held in Brisbane on the 2nd - 4th November 2006 inclusive.**

The international faculty for the meeting includes many of the world's foremost experts in areas including psychiatry, speech and language, cardiology, molecular genetics, clinical genetics, education, feeding, reconstructive surgery and intellectual performance. There are also presentations by VCFS sufferers or their carers which have been proven popular in past meetings.

Some of the speakers there will be:-
Dr. Shprintzen who is the world's foremost authority on this complex, perplexing and surprisingly common genetic disorder.
Dr Karen Golding-Kushner (author of the pre-eminent text in speech therapy for VCFS patients) will be presenting a paper.

The faculty will also offer brief private consultations for children and adults with VCFS to address concerns such as speech disorders, feeding disorders, leg pains and behavioral problems.

Social Events and what is new this year are also on the agenda.

You can go to our website for more information www.vcfs.com.au.
Please contact us with any queries and look forward to hearing from you.
Kathy Russell
VCFS Foundation
1 Milman Street
Clayfield Qld 4011
Email krussell@vcfs.com.au
Web www.vcfs.com.au

## Klinefelter Support Group Meeting – Saturday 12th October 2006

The Klinefelter Support Group meeting
Saturday 21th October at 2.00pm
Jika Jika Community Centre (
Cnr of Plant and Union St Northcote,
Melways Map 30 F11) .

This meeting is to discuss what people want from the group. Partners are most welcome to attend as Klinefelters affects the whole family - not just one person. To RSVP, e-mail kieran215@hotmail.com or pbynemoroney@yahoo.com.au, or call Kieran on 0419-556-825 (please leave a message) or 9248-8419 (after 2pm).

## Kabuki Syndrome Family Day – Saturday 28th October

Noah's Ark Family Resource Centre, 37 Fenwick St, Geelong.
Arrival time is 11.30 am to 12
Lunch 12.30
Speakers 2.00 pm.
Geneticist Dr Sue White.
Please contact Darrin or Stacey on 03 5275 1542, or 0408 552 914, or by email on dsmckiernan@dodo.com.au .

**Smith-Magenis Syndrome Support Group**
Symptoms of Smith-Magenis Syndrome may include developmental delay, intellectual disability, low muscle tone and behavioural issues. Smith-Magenis Syndrome Australia organises regular meetings for families and provides an opportunity to speak with other people in similar situations. People affected by this condition are invited to Chris Blanchard
Email: cblanchard@csu.edu.au

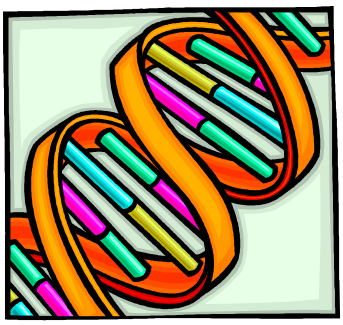
## Microcephaly Support Group

If anyone would like more information or are trying to find a support group NSW please contact
Merey Turner on 02 66822173.

# OVERSEAS NEWS

## National Tay-Sachs & Allied Diseases Association (NTSAD)

29th Annual Family Conference: April 19-22, at the Quincy Marriott in Quincy, Massachusetts.
For more information visit www.ntsad.org or call 800-906-8723.



# A – Z of GENETIC CONDITIONS

*It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness without our membership of the range of genetic condition. 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 impotence that you let us know your group is "Alive and Well' and happy to take referrals.*

*We are happy to profile Kabuki syndrome and we kindly thank the Centre for Genetics Education for their fact sheet.*

# *KABUKI SYNDROME*

## *ALSO KNOWN AS*

- KABUKI MAKE-UP SYNDROME
- NIIKAWA-KUROKI SYNDROME

## *GENERAL INFORMATION ABOUT KABUKI SYNDROME*

Doctors in Japan first described kabuki syndrome in 1980. The name "Kabuki" was selected because of the facial resemblance to the make up of actors in Kabuki traditional Japanese theatre.

Kabuki syndrome is a rare condition with the following characteristics:

- Intellectual disability;
- short stature;
- unusual facial features;
- abnormalities of the skeleton;
- unusual skin ridge patterns on the fingers, toes, and palms of the hands and soles of the feet.

The majority of the reported cases of this condition have occurred for no apparent reason (*sporadic*). However several cases have been reported to be inherited in families.

The most distinguishing features of Kabuki syndrome are the facial features. The following is a list of these features:

- Long palpebral fissures
- Long/ thick eyelashes
- High/ arched eyebrows
- Cleft or high arched palate
- Lowset/ prominent ears
- Broad and depressed nose tip.

Many patients with Kabuki syndrome have a sideways curvature of the spine called *scoliosis*, a short fifth finger that curves inward, and abnormalities of the vertebrae, hands and hip joint. Short stature and abnormal skin ridge patterns on the fingers, toes, palms of the hands and soles of the feet are also common.

Most children with Kabuki syndrome have a mild to moderate intellectual delay. A few, though **they may** need assistance with speech and fine motor skills, are able to follow the regular curriculum in school

Many of the older children (in their early teens) have learned or are learning to read at a functional level. Math skills vary; some do fairly well, while others continue to struggle with this. Over 50% of people with Kabuki syndrome experience hearing loss. The loss in most cases, however, does not result from repeated ear infections (implying damage to the ear).

Most people with Kabuki syndrome have joint laxity. Hip abnormalities, resulting in hip displacements, are a possibility. There is also a possibility of dislocation of the patella (kneecap), most likely arising because of lax ligaments. At least 30% of the children with Kabuki have heart defects. For most, this is discovered soon after birth and corrected by surgery. A significant amount of the children have kidney or other urinary tract anomalies.

There are many other manifestations being reported such as wide set nipples, premature breast development, early puberty, lowered immunity, microcephaly, undescended testes, umbilical hernias, inguinal hernias, generalised hirsutism (hairiness) and vitiligo vulgaris (patches of de-pigmentation).

Not all children manifest all these characteristics. The phenotype or physical characteristics of the condition seem to evolve over time making diagnosis in infancy difficult sometimes.

## *WHAT CAUSES KABUKI SYNDROME?*

The majority of cases of Kabuki syndrome are thought to occur for no apparent reason (*sporadic*). There have been a few reports in the medical literature of families in which the condition appears to be inherited, following a pattern called autosomal dominant inheritance. In these families, it is found that the expression or the symptoms amongst affected family members may vary greatly. This is called variable expressivity.

*For more information about genes, chromosomes and the autosomal dominant form of genetic inheritance, please refer to Genetics Fact Sheets 1, 3 and 7.*

### *WHO IS AFFECTED BY KABUKI SYNDROME?*

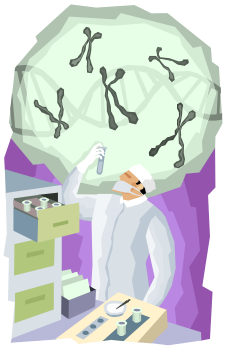
Kabuki syndrome is a rare condition that affects males and females in equal numbers. The majority of cases have been reported in people of Japanese ancestry; however, it is still probably greatly under-diagnosed since the number of medical professionals who are familiar with the syndrome is still growing. Diagnosis is further complicated by the fact that the spectrum of "symptoms" is very diverse. As more and more geneticists become aware of Kabuki syndrome, more children are being diagnosed.

### *IS THERE ANY TREATMENT FOR KABUKI SYNDROME?*

People with Kabuki syndrome may benefit from orthopaedic care and physical therapy for prevention of skeletal problems. Cosmetic surgery may also be helpful for some features. Treatment of biliary, respiratory problems or other problems is symptomatic and supportive.
Genetic counselling may be of benefit to individuals with Kabuki syndrome and their family.

**The following Internet Network Group may be able to provide additional information and support:**

**Kabuki Syndrome Network**
Home Page: http://www.kabukisyndrome.com/



# *Our Little Man has Kabuki Syndrome*.

At 10 weeks I discovered I was pregnant with twins, I remember spending the next hour laughing from shock. The following weeks were the usual twin pregnancy morning sickness, cravings, and anticipation of an expectant mother. At 19 weeks a routine ultrasound showed that twin 2 had a cleft lip and pleural effusion; our doctor broke the news gently and then told us that the baby had a good chance of being Down syndrome. I was devastated. We asked the doctor for an amniocentesis and he performed it the next day; we were then informed that we would have to wait up to a month for the results. Time stood still, and an overwhelming feeling took over our lives. After two weeks we began daily telephone calls to our doctor to see if the results were in, and at day 21 the receptionist told us the results had arrived but our doctor was away, and she couldn't give the results to us. My husband jumped into the car and headed for the surgery. Once there the receptionist said that the notes were on the desk and she would look away; I guess the sight of a man with tears in his eyes were too much for her to ignore. He read the results and came home; he was crying and smiling at the same time, I knew the news was good. I spent the next few weeks very uncomfortable but content knowing that my fears of having a child with Down syndrome weren't an issue any more.

My next appointment was with a different doctor because mine was away, he seemed a little concerned about the growth of my tummy, I was 21

weeks. I assumed my little bubs were content and growing well. While we were leaving he asked me to go and have an ultra sound ASAP; and the next available appointment was for the next day. This ultra sound took 2½ hours and eventually the lady performing the scan said "I am not supposed to say anything, but your baby is very sick" they (the radiographer) told me to wait while they rang the doctor; who told my husband over the phone to get me to the WCH as soon as possible. So much inside you just shuts down. Like a feeling of numbness. On arrival we were shown to a suite where the bustle of nurses was just a dull noise as my heart was breaking, our doctor told us it looked very grave. I had nothing to say to any one including my husband. A spinal block was inserted and the Caesarean Section commenced; twin 1, our little girl Hannah was born first and I couldn't believe how small she was, then twin 2 next. I so vividly remember laying there wishing to run after the babies that they had pulled from my body, but I couldn't move, instead I lay listening and anticipating the cries of new born babies, but they didn't come. The next thing I remember was incredible pain as the spinal block failed and they knocked me out. When I woke I remember them fussing about my blood pressure when all I wanted was to find my babies. Hours later they offered to take me to NICU so I could see Hannah and Zachary; I looked at Hannah she was small but I wasn't worried. Then I met Zachary…

I can't believe the multitude of tubes and monitors. A very bloated baby that looked incredibly ill. This was my son. The nurses explained that Zachary had a condition called hydrops fetalis which simply means his little premature body was full of fluid.

The next hours, days, months and years tumble weeded into a myriad of life threatening illnesses, operations, physical and mental disabilities and discovery. Some of the features included:

NECH (necrotising enterocolitis is an acquired disease, primarily in premmies or sick babies in which intestinal tissue dies.)
Bilateral hernias: (simultaneous right and left inguinal hernia.)
Undescended testicle
Cranial Haematoma
Plural effusion
Coarctation of the aorta: (a congenital constriction of the Aorta, impeding the flow of blood below the level of the constriction and increasing blood pressure above the constriction.).
Bi cuspid valve: (due to a congenital deformity. A normal aortic valve has three cusps, where a bicuspid valve has only two.
Aortic stenosis
Cleft lip
Cleft palate
Ear infections
Nephrocalcinosis
Kidney stones
Eptopic kidney
Cysts
Bronchi pulmonary dysplasia
Acute jaundice

His PHYSICAL FEATURES included:-
Unusual eyes
Sacral dimple
Lip pit
Double jointed
Low muscle tone
Strabismus
Short stature
Oral sensitivity
Tactile sensitivity
Long palpebral fissures
Arched eyebrows
Long eyelashes
Prominent ears
Depressed nasal tip

It is with this experience that I can smugly say…I, now know the true meaning of life and love.

When we found out that Zachary has Kabuki Syndrome it was very hard, even though it all made sense we had to grieve for the son we thought we had and would never have.

Zachary has three sisters; they love him just as they are supposed to. He is in a mainstream school with support, and I truly believe that they are richer for knowing him.

Our children come to us all different; they need our love and guidance from the day they are born until the day we die, cherish them.

Our website is www.sakks.org

By Peta Colton

# LIVING WITH EPILEPSY?

Epilepsy Action is unveiling plans to improve the quality and reach of our education and support services for Australians with epilepsy and other seizure disorders.
www.epilepsy.org.au

When: 11am – 1.00 pm
Tuesday 17th October 006
Venue: The Metcalfe Auditorium, Grd Flr
Mitchell Wing, State Library of NSW,
Macquarie Street, Sydney

Speaker: Epilepsy Action CEO Carol Ireland

*Includes refreshments and light lunch*
**This is your opportunity to:**

- Hear more about exciting plans to enhance epilepsy services
- Register your interest to get involved in consumer research into the needs of people living with epilepsy
- Become an Epilepsy Action member and have your say in our future direction

If you wish to attend, **please RSVP by 1 October 2007** so we know numbers for catering. For more information and to RSVP to attend the Annual General Meeting, contact Rhonda Brennan on (02) 9856 7088 or rbrennan@epilepsy.org.au

## *An exciting new development in the Pacific*

It is with great pride and pleasure that the Board of Directors of Australia Pacific Islands Disability Support (APIDS) - Daniel Stubbs, William Jolley, Maryanne Diamond, Deborah Rhodes and Robyn James - announce their first membership and fund raising drive.

APIDS is a not for profit company that is seeking to work with disabled peoples' organisations to improve the lives of people with disabilities in the Cook Islands, Federated States of Micronesia, Fiji, Kiribati, Nauru, Niue, Palau, Papua New Guinea, Republic of the Marshall Islands, Samoa, Solomon Islands, Tonga, Tuvalu and Vanuatu. These are the island member states of the South Pacific Forum.

Joining as a member of APIDS for $20 will make a significant difference to disabled peoples' organisations who work hard in their countries to assist all people with disabilities to have a better quality of life. This includes enough food to eat, the chance of obtaining a job, obtaining the necessary equipment to get around.

An example of what impact your $20 will make - in Fiji, $20 will pay the salary and expenses of one day for the Advocacy Officer with Fiji Disabled Peoples Association.

Thank you for considering this request - APIDS would love to have you join an organisation which has the simple purpose of supporting people with disabilities in the Pacific Islands to fulfil their dreams.

Please note that supporters in Tasmania, Victoria, South Australia, Northern Territory and the Australian Capital Territory can donate using credit cards through the APIDS page on the Our Community website. APIDS is still working on permits in other states.

Contact Robyn James for more information on 0410 085 140, or email apids@aapt.net.au.

# Jasmine's Journey

This is the story of Jasmine's journey – it's a tough road for someone so young to have traveled. It is a story about our search for some answers to her very rare genetic condition. Jasmine recently turned two and we really celebrated this birthday, as for some time we thought it might be a day she would not live to see.

During my pregnancy I had numerous ultrasounds and we were watched carefully, as Jasmine did not seem to be growing as she should be. Jasmine was born at 4 pounds 12 ounces (2170 grams) and was diagnosed with IUGR (Inter Uterine Growth Retardation). Jasmine stayed in the hospital for three weeks and had some tests due to a large fontanel and projectile vomiting. These tests included chromosomal studies, and abdominal and fontanel ultrasounds. When we were allowed to take her home the doctors weren't overly concerned so neither were we – we thought she just needed a bit of a growth spurt and everything would be "fine".

However, six months later Jasmine's height was just 57 cm and she only weighed in at 4.3 kg. Although this was showing appropriate growth along her growth curve, it was so far below her chronological age for weight and height we thought it was time to start a search for some answers. Her doctor suspected "silent reflux", as she was still vomiting a lot but without discomfort. She had a barium swallow done as well as blood tests for Growth Hormone Deficiency and chromosomal studies. These tests all came back normal. We then saw a dietitian who suggested thickening Jasmine's formula and adding extra calories to her meals in an attempt to help control the vomiting and put on some weight. This proved unsuccessful. The vomiting continued and there was very little weight gain.

At nine months old, Jasmine weighed 5.32 kg and was 60 cm tall. Our next step was a referral to the Sydney Children's Hospital Genetics Department. The waiting list was long, and Jasmine finally went to see them when she was eleven months old. It was there that we hoped we would find our answers. The geneticists were confident that Jasmine had a genetic condition. But which one?
This is where more tests started. Jasmine had a skeletal x-ray and a bone age study done.

at 4 months

Jasmine's Naming Day, Oct 2004

Over the next few months, Jasmine was clinically diagnosed with possible Russell-Silver Syndrome (RSS) and a DNA study was done to confirm this diagnosis. While we were waiting for confirmation, naturally we researched into RSS and realized that we needed to get Jasmine to an endocrinologist and a dietitian. With help from a family friend we did that. We were introduced to possible Growth Hormone Injections and G-Tube Feeding. We contacted

organizations to try and get as much information as we could. We were told this condition was quite rare. After further investigations, doctors decided that RSS didn't seem to fit all the pieces of the puzzle and Jasmine's DNA studies came back as negative for Russell-Silver. Back to the drawing board.

Jasmine's case was starting to attract quite a lot of attention in medical circles. Her skeletal x-rays had been forwarded to paediatric radiologists at Westmead Children's Hospital. After quite some time, a professor at Westmead suggested another diagnosis: 3M Syndrome. The genetics teams at both hospitals seemed confident that this was the answer. We were told that basically, there was nothing we could do medically to help her. There would be no developmental problems or health issues - Jasmine was always going to be smaller than average. We just had to help her adjust to life like this.

At sixteen months old, Jasmine weighed 6.26 kg and her length was 67 cm. She was developing well, attending daycare and ruling the household! After so many months of medical tests, life for Jasmine and our family seemed to be getting back to normal. Despite everything, our little girl still had a strong will and a BIG character!

In October, 2005 Jasmine's journey took a new and unexpected direction…

We first noticed something was wrong when her stomach became distended and she began having trouble breathing. Jasmine had an abdominal x-ray, which showed she was constipated. We knew she wasn't herself, and treatment wasn't working. Things just weren't right. So on October 4, we took her to the Emergency Department at Sydney Children's Hospital in Randwick. We said to ourselves, "We are not leaving here until we get some answers". Although we were seriously worried, we had no idea that the answer was going to be so serious.

That same day, we were told Jasmine had a tumor in her right kidney, and further tests needed to be done to determine the seriousness and extent of the tumor. We were told to arrange care for our four year old son Jaidyn, as we wouldn't be leaving hospital for some time.

The next day, Jasmine was scheduled for a CT scan, but just before she was to have her scan we received news from the genetics department. Ironically, the tumor had helped them to finally determine Jasmine's genetic condition: Mulibrey Nanism. The CT scan then confirmed that Jasmine had a Stage 4 Wilms Tumour in her right kidney, traveling to her heart, with secondary tumors in her liver and lung. What a day: two diagnoses, neither of which we wanted to hear. This began another path for us to follow, not just with our daughter now having a life threatening illness, but also diagnosed with a different, and unbelievably rare genetic condition.

The following day, on October 6, Jasmine went straight into surgery and had a portacath put in her chest and had her first round of chemotherapy. We were amazed that within 48 hours her wonderful team of doctors had been able to diagnose Jasmine's illness and begin treatment. The doctors urged us to focus on Jasmine's battle ahead, and our confidence in the doctors helped us to do this.

We were then told Jasmine would most likely not be going home for quite a while as she needed to start aggressive chemotherapy; in addition, doctors had found a large mass of tumor near her heart. For safety reasons they decided it was best for her to be in hospital, in case the tumor in her IVC and the mass at her heart broke away and went to her lungs. "We will try and get her home for Christmas" was all we had to go on. We knew the doctors would do everything they could to make that happen.

With her first round of chemotherapy, Jasmine seemed to go well and had minimum side effects. But almost a week later, just a day before her second treatment, she was happily playing and ordering everyone around when she suddenly became quite upset and lethargic. When I placed her on the bed to change her nappy, we found her nappy full of blood. Jasmine was bleeding heavily from somewhere and we really had no idea why. Within fifteen minutes, Jasmine had been examined by her doctors and surgeons, had x-rays done, and had started a blood transfusion. We had thought a lot could happen in one day – now we new how much things could

change in a quarter of an hour! As frightening as this experience was, it also reinforced our confidence in Jasmine's incredible team of doctors and nurses.

They then gave us a probable cause of the bleeding: it seemed the chemotherapy had already started to work and was breaking up the blood vessels in her kidney, which was then bleeding through her urine. So the chemotherapy was working – we didn't realize the results would be quite so immediate, or dramatic. Jasmine was now connected to a catheter and having blood transfusions. The bleeding took about a week to stop completely.

Jasmine and Mum on bandana day

Jasmine continued with chemotherapy treatment weekly, and did quite well for someone her size. She had her good days and her bad days. Unfortunately by week three she needed to be tube fed, as her appetite had gone and her weight was going down. She adjusted to this well. Jasmine was very much running the ward by now and was loved by so many people. It was amazing that such a little girl could have such a huge impact on everyone around her – so many people told us that seeing her happy smiling face was what helped them through the ordeal of having their children in the oncology ward. The hospital had become her home and she loved having all these new faces around her.

By November, Jasmine's surgeon decided she was ready for surgery: the mass near her heart had gone, and the tumors in both the kidney and IVC had reduced sufficiently for them to operate. The doctors told us of the dangers of such a difficult operation, and that Jasmine may need a heart by-pass. We were told to prepare for the worst. On November 24, 2005, we said goodbye to our little girl as she went into the operating theatre. Then, we just waited and prayed. I could honestly say, this was the hardest day I have ever experienced - even after everything else we had already been dealt with. After a seven-hour operation, Jasmine's wonderful surgical team told us she was in the Intensive Care Unit. She had done amazingly well – no heart by-pass was need, and the tumors looked to be all gone!

Jasmine was put in a medically-induced coma for about two days. Those first few hours in the ICU were the hardest as this is when it all hit me. I realized just how sick she was. She looked tiny and frail and helpless, and unable to fight. Up until now, despite the severity of her condition, she had always managed to smile and play and almost seemed to carry on "as normal" – some days it was surprising to realize she was actually really sick! We'd been told that she would probably be in the ICU for a week or more – but not our Jasmine. By day three she was back up in the ward and trying so desperately to get up and go for a walk around.

One week after surgery Jasmine was well enough to start radiation. She had six sessions over two weeks, and through the break time we even got to take our little girl home for a weekend. It was the best feeling - we could actually see an end to our three-month hospital stay! Then we were given the best Christmas present we could ever ask for… Jasmine was discharged from hospital on December 15. Our lengthy stay in hospital had finished and we were home! Jaidyn had his mummy back full-time – not just one night a week - and most importantly, he had his little sister home again to play with and protect.

Jasmine was still being tube-fed at home and going back to hospital every few weeks for chemotherapy. It was a lot of hard work and lots of trips back and forth to hospital, but it was definitely worth it to have Jasmine back home. It was a busy time for us, as Jaidyn was starting "big school", and we were so thankful to be able to be there for him. Little things started to take on a whole new significance for us, and we began to realize that every day comes only once.

We were very lucky - Jasmine only had to go back for a couple of hospital visits with temperatures and a port infection. April was a good month for Jasmine – she had her last chemotherapy treatment, her feeding tube came out, and she had a CT scan and was classed as "in remission"! Then, on May 14, we celebrated her 2nd birthday!! We had a huge party for her and invited a lot of close friends and family to celebrate. It was also a chance for us to thank some of the many people who had shown Jasmine their love and support over the past months. Most importantly, we could show off our little hero! She is now having regular scans and ultrasounds and we are still praying every day that she can stay in remission and keep fighting.


Jasmine off to a clinic visit


Playing at home

With Jasmine now finished treatment, it was time to concentrate on her genetic condition again. We needed to start the search for some answers and to find out how the rest of her life would be affected. We were given a definite diagnosis of Mulibrey Nanism, which seems to be dominant in Finland with 85 of the 113 known cases in the world from there. (Interesting, as neither of us have any Finnish ancestry that we know of.) Mulibrey is an acronym of muscle-liver-brain-eyes, the organs most frequently involved. Nanism is another word for dwarfism. Finally we had a definite answer!

We began researching Mulibrey Nanism. Little did we realize how hard this was really going to be! Our doctor was not even sure what to tell us about this condition because so little is known about it. They could tell us however that it was a very rare condition with Jasmine looking to be the first known case in Australia.

Jasmine presented with the following features associated with Mulibrey Nanism: short stature, enlarged liver, relatively large hands and feet, Wilms Tumor, feeding difficulties, cranio-facial features. We were lucky that she has not yet presented with any possible heart problems such as pericardial constriction.

Unfortunately, out of the 113 cases, Wilms Tumor is a rare feature of Mulibrey - Jasmine is only about the third child to develop the tumor from this genetic condition. She also has cystic malformations on her lung which are being watched, but this actually has nothing to do with her genetic condition. Just an "added bonus", as we say.

We visited the Connective Tissue Dysplasia Clinic at Westmead Hospital, who checked her development – all fine - and gave us advice about a few things we would need to look into further when Jasmine is older, such as getting on the toilet, into bed, and so on. They also gave us a growth chart for Mulibrey Nanism, but we have not yet been able to understand it as it is in Finnish.

Where do we go from here?? We tried contacting the University in Finland where Jasmine's DNA had been sent and we got nowhere there either. We contacted every organization we could find and to this date still have very limited information. We are not sure who we should be seeing, or when we should be seeing them. Nor do we really know how Jasmine will be affected throughout her life by this condition. How do we explain to her when she is older what she has, when we don't really understand it ourselves? How do we give her the hope that she will be able to lead a normal life like her friends, when we don't know? We need to find some answers for us and for Jasmine.

Recently we were lucky enough to get in contact with a family in America who has a 12 year old boy

with Mulibrey Nanism. They have been gracious enough to share their story and their search for answers with us. This has helped and inspired me to write Jasmine's story, in an attempt to get it out to as many people as possible. Hopefully this story will find its way to a medical professional who will want to help us find some answers for our little girl. Hopefully this story might also find its way to parents of a child who may have symptoms similar to Jasmine, and by being aware of this condition will lead to an earlier diagnosis.

So far, Jasmine's journey has been a very challenging one - but hopefully it will continue to lead in the right direction. There's something about Jasmine that just stops people in the streets. It's amazing the effect she has on people. She radiates happiness. She has a giant spirit for such a little girl and is always smiling and full of energy. People often ask my husband and I how we coped through all of this, and we always tell them the same thing - Jasmine is the one that gets us through. If she can smile then we have no right but to smile with her. She is my hero and my princess. All we would like for her now is to get her through each day of her remission process, have her classed as cured in five years' time, and hopefully succeed in our search for answers. After all, this isn't just any search and rescue mission: it's for our Jasmine's health and well being.

Jasmine has been able to start a Physical Culture class

**Kim Clingan**

the family together on a recent holiday to Queensland

## Genetic Disorders Awareness Week - "Adults Bridging the Gap" (26th July 2006)

Debbie, Dianne, Belinda, Sue, David, Peter & Jane


Belinda, Sue, David, Peter, Dianne

Anthony Earp, Charles Adderley, Peter Berner

Lynne Brodie, Anne Cumming, Jennifer Blackwell

Trudy & Phil Whaite, Dr. Kristine Barlow-Stewart

## WAO / IGA

Prof. Arnold Christianson
11th International Congress of Human Genetics
6th August 2006, Brisbane

**Creating awareness and providing contact, peer support and education to people affected by genetic disorders.**

**"MAKING THE RIGHT CONNECTIONS SINCE 1988"**

*Funded by the NSW Health Department*

*You are invited to attend*

# AGSA's

## Annual General Meeting

Sunday 19th November 2006

at 66 Albion St, Surry Hills (Old Children's Court Building)

Time: 11.00 am

## AGENDA

1. Welcome and Introduction
2. Apologies
3. President's Report

Guest Speaker

4. Information Officer's Report
5. Treasurer's Report and audited accounts
6. Election of Office Bearers for 2005/06
   a) All positions declared vacant and appointment of returning officer
   b) Election.
   The committee, including executive, comprises a maximum of 10 members.
7. Appointment of Public Officer
8. Appointment of Auditor
9. General business

10. Close of business

*Light Refreshments will be served*

*If you wish to attend for catering purposes please contact*

**Dianne Petrie:**
**AGSA, 66 Albion Street Surry Hills NSW 2010 Australia**
**Ph: + 61 2 9211 1462 Fax: + 61 2 9211 8077**

**website: www.agsa-geneticsupport.org.au**

**email: dianne@agsa-geneticsupport.org.au**

# ELECTION OF OFFICE BEARERS FOR 2006/07

# NOMINATION FORM

NB Only financial members at the time of the AGM are eligible to vote and/or hold office.

I.....am willing to accept

nomination for the position of.....within the

Association of Genetic Support of Australasia Inc.

Signed.....

Date.....

Proposed by: .....

Signed .....

Date .....

Seconded by: .....

Signed .....

Date .....

# Form of Apppointment of Proxy
# Rule 33 (2)

I, ……
(full name)

of ……...
(address)

being a member of the Association of Genetic Support of Australasia Inc, (AGSA) hereby appoint

……

of……
(full name of proxy)

being member of that incorporated association, as my proxy to vote for me on my behalf at the Annual General Meeting of the Association to be held on Sunday the nineteenth day of November 2006.

……..
Signature of member appointing proxy

……..
Date

## List of Conditions on AGSA's Contact Register

*Aarskog syndrome*
*Achondrogenesis*
*Achondroplasia*
*Acoustic Neuroma*
*Acrocallosal syndrome*
*Acromegaly*
*Adams Oliver syndrome*
*Addison's*
*Adrenoleukodystrophy*
*Agenesis of the Corpus Callosum*
*Alagille syndrome*
*Albinism*
*Alcardi syndrome*
*Alexander Disease*
*Alpha Manniosidosis*
*Alpha1 Antitrypsin Deficiency*
*Alpha Thalassaemia X-Linked Mental Retardation*
*Alport syndrome*
*Alstroms syndrome*
*Amyloidosis*
*Amyotrophic Lateral Sclerosis*
*Angelman syndrome*
*Aniridia*
*Ankylosing Spondylitis*
*Anodentia ñ Congenital*
*Anticardiolipin AB Type*
*Anti-Phospholipid Syndrome*
*Apert syndrome*
*Aplasia Cutis Congenita*
*Argininosuccinic*
*Aciduria & Citrullinaemia*
*Arnold-Chiari*
*Arthrogryposis*
*Aspergers syndrome*
*ATR16 syndrome*
*Autism*
*Baller-Gerold syndrome*
*Bannayan-Riley-Ruvalcaba syndrome*
*Bardet-Biedl syndrome*
*Bartter syndrome*
*Basal Nevus syndrome*
*Batten Disease*
*Beckwith-Wiedemann syndrome*
*Behcetís syndrome*
*Behr syndrome*
*Benign Essential Tremor*
*Berardinelli syndrome*
*Bilateral Iris Coloboma*
*Binder syndrome*
*Bloom syndrome*
*Blount's Disease*
*Borjeson-Forssman-Lehmann syndrome*
*Brown syndrome*
*BRCA1/2*
*Caffeys familial neurovisceral lipidosis*
*Caffeys generalized gangliosidosis*
*Caffeys Pseudo -Hurler syndrome*
*CAH & Hypoplasia Duchene Muscular Dystrophy*
*Camptomelic Dysplasia*
*Canavans Disease*
*Cardio facial cutaneous syndrome*
*Cardiomyopathy*
*Caroli syndrome*
*Carpal-tarsal osteolysis*
*Carpal Tunnel syndrome*
*Carpenter syndrome*
*Central Core Disease*
*Cerebellar Hyperplasia*
*Cerebo-Costo-mandibular*
*Charcot-Marie-Tooth Disease*
*Choanal Atresia*
*Chromosome 18q deletion*
*Chromosome 18p deletion*
*Chronic Granulomatous Disease*
*Choroid Plexucyst*
*Chronic Granulomatous disease*
*Cleidocranial dysplasia*
*Cobalamin E, C/G deficiency*
*Cockayne syndrome*
*Coffin-Lowry syndrome*
*Coffin-Siris syndrome*
*Cohen syndrome*
*Congenital Adrenal Hyperplasia*
*Congenital Alopecia Totalis*
*Congenital Anodontia*
*Congenital Cone dystrophy*
*Congenital Fibre Type Disproportion*
*Congenital Myotonia Dystrophy*
*Congenital Protein C deficiency*
*Conradi-Hunnermannís*
*Cornelia-de Lange syndrome*
*Corticobasal degeneration*
*Costello syndrome*
*Craniosynostosis syndrome*
*Cri-du-chat syndrome*
*Crouzon syndrome*
*Cushing syndrome*
*Cutis Marmorata Telangiectatica*
*Cyclical Vomiting Syndrome*
*Cystic Fibrosis*
*Cystinuria*
*Cytochrome C. Oxidase Deficiency*
*Dancing Eye syndrome*
*Dandy-Walker Malformation*
*De Barsy syndrome*
*Dejerine-Sottas disease*
*Dercum Disease*
*Desbuquois syndrome*
*Developmental Verbal Dyspraxia*
*Diastematomyelia*
*Di-George*
*D 2 hydroxyglutaric aciduria*
*Drash syndrome*
*Double Y syndrome*
*Duane syndrome*
*Dubowitz syndrome*
*Dysautonomia*
*Dyschondrosteosis*
*Ebsteins Anomaly of the Tricuspid Valve*
*Ectodermal dysplasia*
*Ectrodactyly*
*Ehlers Danlos syndrome*
*Ellis-Van Creveld Syndrome*
*Emery Dreifuss Muscular Dystrophy*
*Encephalocraniocutaneous Lipomatosis*
*Epidermolysis Bullosa*
*Epidermal nevus syndrome*
*Erythropoietic protoporphyria*
*Fabrys Disease*
*Facial Haemangioma*
*Factor V Leiden*
*Familial adenomatous polyposis coli*
*Familial Hiberian Fever*
*Familial Hyperinsulinaemia*
*Familial Mediterranean Fever*
*Familial Spastic Paraparesis*
*Fanconi Anaemia*
*Farber Lipogranulomatosis*
*Fazio-Londes syndrome*
*FG Syndrome*
*Fibrodysplasia Ossificans*
*Fish Odor syndrome*
*48, XXXY*
*48, XXYY*
*49, XXXXY*
*Fragile X syndrome*
*Fraser syndrome*
*Friedreichís Ataxia*
*Froelich syndrome*
*Frontanasal Dysplasia*
*Fryns syndrome*
*Fukuyama syndrome*
*Galactosaemia*
*Gardner syndrome*
*Gastroschisis*
*Gaucher Disease*
*Gitelman syndrome*
*Glucose 6 Phosphate Dehydrogenase (G6PD) deficiency*
*Glucosidosis Enzyme deficiency*
*Glycogen Storage Disease*
*GM1 Ganliosidosis*
*Goldenhar syndrome*
*Gorlin syndrome*
*Graves Disease*
*Guillain Barr syndrome*
*Haemochromatosis*
*Hailey-Hailey Disease*
*Hajdu-Cheney syndrome*
*Hallermann-Streiff syndrome*
*Hallervorden-Spatz Disease*
*Hartnup Disease*
*Hemihypertrophy*
*Hemimegalencephaly*
*Hereditary Angioneurotic Edema*
*Hereditary Fructose Intolerance*
*Hereditary Haemorrhagic Telangiectasia (Rendu Osler Weber syndrome)*
*Hereditary Multiple Exostoses*
*Hereditary non polyposis colorectal cancer*
*Hereditary Spastic Paraplegia*
*Hereditary Spherocytosis*
*Hirschsprungís Disease*
*Holoproscenphaly*
*Holt Oram syndrome*
*Holoprosencephaly*
*Homocystinuria*
*Homolateral Brain syndrome*
*Hunter syndrome*
*Huntington Disease*
*Hydrocephalus*
*Hydronephrosis*
*Hyperargininaemia*
*Hyper IGE syndrome*
*Hyperexplexia (Startle Disease)*
*Hypertrophic Cardiomyopathy*
*Hpoochondroplasia*
*Hypoplasia of the Cerebellum*
*Hypomelanosis of Ito*
*Hypophosphatemic Bone Disease (HBD)*
*Hypophosphatasia*
*Hypopituitarism*
*Hypoplastic left heart syndrome*
*Hypoplastic Primary Vitreous*
*Hypotension Orthostatic*
*Hypothyroidism*
*Idiopathic pulmonary fibrosis*
*Idiopathic Thrombocytopenic Purpura*
*Immotile Cilia syndrome*
*Incontinentia Pigmenti*
*Ivermark syndrome*
*Jacobson syndrome*
*Jeune syndrome*
*Job syndrome*
*Johanson-Blizzard syndrome*
*Joubert syndrome*
*Kabuki Make-up syndrome*
*Kallmann syndrome*
*Kawasaki syndrome*
*Kearns Sayre syndrome*
*Kennedyís disease*
*Keratosis follicularis spinulosa decalvans*
*Klinefelter syndrome (47,XXY)*
*Klippel-Feil syndrome*
*Klippel-Trenaunay Weber syndrome*
*Krabbe Disease*
*Kyphomelic Dysplasia*
*Landau-Kleffner syndrome*
*Langer-Giedion syndrome*
*Larsen syndrome*
*Laurence-Moon-Biedl*
*Lebers Optic Atrophy*
*Leigh Disease*
*Leopard syndrome*
*Leri-Weill syndrome*
*Lesch Nyhan syndrome*
*Leukodystrophy*
*Li-Fraumeni*
*Limb Girdle Muscular Dystrophy*
*Lipodystophy & Brownís syndrome*
*Lissencephaly*
*Long-chain-3-hydroxyacyl coenzyme A, dehydrogenase deficiency*
*Long QT syndrome*
*Loweís syndrome*
*Lujan-Fryns syndrome*
*Lymphas Genphasia*
*LAM (lymphangioleiomyomatosis)*
*Lysosomal Storage Disorders*
*Machado Joseph syndrome*
*Maple Syrup Urine Disease*
*Marfan syndrome*
*McCune Albright (Polkystotic Fibrous Dyplasia)*
*McKusik Kaufman*
*Megalocornea Mental Retardation*
*MELAS syndrome*
*Menke syndrome*
*MEN2*
*Metachondromatosis*
*Metatropic Skeletal Dysplasia*
*Methylmalonic academia*
*Microcephaly*
*Miller-Dieker syndrome*
*Minicore disease*
*Mitochondrial Myopathies*
*Moebius syndrome*
*Monosomy 9p*
*Motor Neurone Disease*
*Mucopolysaccharidoses*
*Mullerian Duct Agenesis*
*Multiple Endocrine Neoplasia 2B*
*Multiple Epiphyseal Dysplasia*
*Multiple Exostores*
*Muscular Dystrophy*
*Myasthenia Gravis*
*Myotonia Congenita*
*Myotonic dystrophy*
*Nager & Miller syndrome*
*Nail Patella syndrome*
*Narcolepsy*
*Netherton syndrome*
*Neuroaxonal Dystrophia*
*Neurofibromatosis*
*NF + Noonan syndrome*
*Neuronal Intestinal Dysplasia*
*NF1 Noonan syndrome*
*Niemann-Pick Disease*
*Nonketotic Hyperglycinaemia*
*Noonan syndrome*
*Norrie syndrome*
*Nystagmus*
*Oculo-dento-digital syndrome*
*Ohdo syndrome*
*Olivo-Ponto-Cerebellar-Atrophy*
*Olliers Disease*
*Ophthalmia: Anophthalmia & Microphthalmia*
*Opitz Fg syndrome*
*Opitz trigonocephaly*
*Oral-Facial-Digital syndrome*
*Organic acidemia*
*Ornithone transcarbamylase deficiency*
*Osteogenesis Imperfecta*
*Osteopetrosis*
*Ovarian cancer*
*Pachyonychia congenita*
*Paget disease*
*Pallister-Hall syndrome*
*Pallister-Kellian syndrome*
*Paroxysmal Nocturnal Haemoglobinuna*
*Paroxysmal kinesigenic choreoathetosis*
*Partington syndrome*
*Peho syndrome*
*Pelizaeus-Merzbacher Disease*
*Pena-Shokeir syndrome type1*
*Pendred syndrome*
*Peripheral Neuropathy (CMT TypeII)*
*Persistent hyperinsulinenuic hypoglycemia (PHHI)*
*Perthes syndrome*
*Peutz-Jeghers syndrome*
*Pfeiffer syndrome*
*Phenylketonuria (PKU)*
*Picks Disease*
*Pierre Robin syndrome*
*Poland syndrome*
*Polyostic Fibrous Dysplasia*
*Polycystic Kidney Disease*
*Polycystic Ovarian syndrome*
*Pompe disease*
*Popliteal Pterygium syndrome*
*Porphyria*
*Post Polio syndrome*
*Potter syndrome*
*Prader-Willi syndrome*
*Primary agammaglobulinaemias*
*Primary Immune Deficiency*
*Progeria syndrome*
*Progressive Myoclonic Epilepsy*
*Progressive Supranuclear Palsy*
*Proteus syndrome*
*Pseudohypoparathyroidism*
*Pseudoxanthoma Elasticum (PXE)*
*Pycnodysostosis*
*Pyridoxine dependency*
*Pyruvate dehydrogenase deficiency*
*Pyruvatekinase deficiency*
*Rare Chromosomes Disorders ñ includes deletions, inversions, trisomies, duplications, ring, uniparental disomy, mosaicism, tetrasomies, translocations*
*Raynaud syndrome*
*Reinfenstein syndrome*
*Retinitis Pigmentosis*
*Rett syndrome*
*Richardson-Steele-Oblizewski syndrome*
*Ring 18*
*Robinow syndrome*
*Rubenstein-Tabi*
*Russel-Silver syndrome*
*Refsum disease*
*Raynaud's syndrome*
*Saethre-Chotzen syndrome*
*Sandhoffs disease*
*Sanfilippo disease*
*Sarcoidosis*
*Schinzel Giedion syndrome*
*Schmid Type Metaphyseal Chondrodysplasia*
*Schwachman syndrome*
*Septo-optic Dysplasia*
*Severe Combined Immune Deficiency*
*Severe Immune Deficiency*
*Short stature and Skeletal Dysplasia*
*Shprintzen syndrome*
*Shy Drager syndrome*
*Simpson Golabi syndrome*
*Sjogren syndrome*
*Smith-Magenis syndrome*
*Sotos syndrome*
*Spina Bifida*
*Spinal Muscular Atrophy*
*Spinocerebellar Ataxia Type II*
*Spondylocostal Dysplasia*
*Spondylometaepiphyseal dysplasia*
*Sponylo Epiphyseal Dysplasia*
*Shprintzen syndrome (Velo Facial Cardio syndrome)*
*Steve Johnson syndrome*
*Stickler syndrome*
*Sturge Weber syndrome*
*Systemic Lupus Erythematosus*
*TAR syndrome*
*Tay Sachs disease*
*Tetrasomy 18p*
*Thalassaemia*
*Tibial Hemimelia*
*Tourette syndrome*
*Townes Brock syndrome*
*Treacher-Collins syndrome*
*Trichothiodystrophy*
*Trimethylaminuria*
*Triple X syndrome (47,XXX)*
*Trisomy4,5,8,9,10,12,13,18*
*Trisomy/Partial Trisomy*
*Tuberous Sclerosis*
*Turner syndrome (45,X)*
*Tyrosine Anaemia*
*Undiagnosed conditions group*
*Usher syndrome*
*Vater syndrome*
*Velo-Cardio-Facial Syndrome*
*Vitiligo*
*Von Hippel-Lindau syndrome*
*Von Witterbrand disease*
*Weaver syndrome*
*Weill-Marchesani syndrome*
*Werding-Hoffman syndrome*
*West syndrome*
*Whistling face syndrome*
*Wiedemann-Rautenstrauch syndrome*
*William syndrome*
*Wishott-Aldrich syndrome*
*Wolf-Hirschhorn syndrome*
*Wolfrau syndrome*
*Wolman's disease*
*Xeroderma Pigmentosum*
*X-linked Agammaglobulinaemia*
*X-linked Hypophosphatemia (XLH)*
*XLP syndrome*
*Zellweger syndrome*

*PLUS 134 ORGANISATIONS AND SUPPORT GROUP MEMBERS.*

*The views expressed in this newsletter are not necessarily those of AGSA*

**AGSA'S SUPPORT GROUP & ORGANISATIONAL MEMBERS as at January 2004**

Act Muscular Dystrophy Association Inc.
Androgen Insensitivity Assoc. Support Group of Australia
Alagille Syndrome Support Group
Albino Support Group
Angelman Syndrome Assoc. Inc.
Alzheimer's Assoc of Aust Inc.
A.P.I.A. (Aust.Primary Immune Deficiencies Assoc.)
Assoc. for Children With a Disability, Vic.
Assoc. for the Welfare of Child Health (AWCH)
AUSSIE FOLKS
Australian Addison's Disease Assoc. Inc.
Aust. Arthrogryposis Group (TAAG) Inc.
Australian Assoc. for the Welfare of Child Health (AWCH)
Aust. CHARGE Association
Aust. Crohn's & Colitis Assoc.
Aust. Huntington's Disease Association (Qld) Inc.
Aust. Huntington's Disease Assoc. (NSW) Inc.
Aust. Speak East Assoc.
Australasian Tuberous Sclerosis Society Inc.
Aust. Leukodystrophy Support Group
Aust. Society for Ectodermal Dysplasia
Autistic Assoc. of NSW
Batten's Disease Support & Research Assoc. Inc. (Australian Chapter)
Beckwith-Weidemann Syndrome Support Group
Bunyip Special Needs Group Inc.
Cardiomyopathy Assoc of Aust. Ltd.
Centacare Early Intervention.
Centre for Developmental Disability Studies
Charcot Marie-Tooth Assoc. of Australia Inc.
Charcot Marie-Tooth Disease, USA
Child & Family Health Centre
Child Health Information Centre
Community Resource Team (Albury)
CONTACT A FAMILY U.K.
Cleft Pals, The Cleft Palate & Lip Society
CLIMB Children Living with Inherited Metabolic Diseases
Coeliac Society of NSW Inc.
Congenital Adrenal Hyperplasia Support Group
Cornelia de Lange Syndrome Support Group
Cri du Chat Syndrome Support group of Australia Inc.
CVS Support Group (WA)
Cystic Fibrosis Assoc of Qld Ltd.
Cystic Fibrosis Assoc. of Vic
Cystic Fibrosis New South Wales
Early Education Clinic, North Sydney
Early childhood Intervention Program
DIAL (Qld)
Donor Conception Support Group
Depressive & Manic Depressive Assoc.
Dystrophic Epidermolysis Bullosa Research Association (DEBRA) NSW. Inc.
Early Learning Tasmania
Ehlers-Danlos Syndrome Support Group
Exceptional Parent (USA)
Fabry's Support Group Inc.
Family Advocacy
Family Planning Assoc.
Fragile X Assoc of Australia
Friedreich Ataxia Assoc of NSW
Gaucher Assoc. of Australia
Genetic Alliance (USA)
Genzyme Australia Pty. Ltd.
Genetic Interest Group (GIG) (UK)
I.D.E.A.S. Inc
Kidney Kids Support Group NZ
Klinefelter Syndrome Support Group
Kurrajong Early Intervention
Haemochromatosis Society Inc.
Haemophilia Foundation NSW
Hereditary Cancer Registers (NSW Cancer Council)
Hereditary Haemorrhagic Telangiectasia
Hereditary Fructose Intolerance
Hunter Orthopaedia School
IDEAS Inc.
Kidney Kids of NZ Support Group
Maternity Alliance
NALAG
Leukodystrophy Foundation (USA)
Leigh's Disease Support Group
Lowe's Syndrome Assoc. Inc. (USA)
Lower Nth Shore Community Support Team
Lupus Association of NSW Inc.
Lysosomal Diseases Australia
M.P.S. Society
Marfan Syndrome Support Assoc. NSW
Marfan Syndrome Assoc. Australia (S.A.Branch))
Meniere's (NSW) Support Group
Mental Illness Nervous Disorders Association
Metabolic Dietary Disorders Association (MDDA)
Mid North Coast Area Health Taree Genetics Service
Motor Neurone Disease Assoc. of NSW Inc.
Multiple Epiphyseal Dysplasia Assoc.
Muscular Dystrophy Assoc of NSW
Muscular Dystrophy Assoc (NZ) Inc.
National Council of Intellectual Disability
NCOSS (NSW Council of Social Services)
Neurofibromatosis Assoc.
Noonan Syndrome Support Group
NSW Genetics Education Program
NSW Cancer Council
Osteopetrosis Support Group
Osteogenesis Imperfecta of Aust.
Parents Bereavement Support Group
Parent to Parent (NZ)
Pen-Parents of Aust. (ACT)
PKU Assoc of NSW
Polycystic Kidney Disease Association
Psoriasis Society
Pseudohypoparathyroidism Support Group
Pseudoxanthoma Elasticum Support Group
Prader-Willi Syndrome Assoc. of NSW (Aust) Inc.
Pyruvate dehydrogenase deficiency.
Rare Chromosomes Disorders Support Group
Retina Australia (NSW) Inc.
Rett Syndrome Assoc. of Aust.
Royal Blind Society of NSW
SAFDA (Support After Foetal Diagnosis of Abnormality)
SANDS
Short Statured People of Northern Qld
Short Statured People of Aust (NSW)
Short Statured People of Aust (Vic)
Short Statured People of Aust. (SA)
Spinal Muscular Atrophy
Schizophrenia Fellowship NZ
Smith Magenis Syndrome Support Group Inc.
Spastic Society of Victoria
Spina Bifida Assoc. of NSW
Spina Bifida Assoc. of WA Inc.
Society of Ectodermal Dysplasia
Southern Child Care Support Program
Sotos Syndrome Support Group
Steele Street Early Special Education Centre Devonport
St Paul's Special School
The Chromosome 18 Registry & Research Society
The Northcott Society
The Toybox Centre Inc.
Thalassaemia Society of NSW
Turner Syndrome Assoc of Aust. Ltd. (QLD)
Turner Syndrome Assoc of Aust. Ltd. (SA)
Turner Syndrome Assoc. of Aust. Ltd. (NSW)
Uncontrolled Epilepsy Support Assoc (Vic)
United Leukodystrophy Foundation (USA)
Velo-Cardio-Facial Syndrome Foundation of Australia.
Wellington Huntington's Disease Assoc. (Inc.) (NZ)
Western Institute for Self Help (W.I.S.H)
West Syndrome Support Group
Wolf-Hirschhorn 4p- Syndrome Support Group
Williams Syndrome Association of Aust. Inc.

**(NB: This list represents support groups and associations members only. In addition to this list of members AGSA has established a Contact Register over 550 genetic conditions representing families and individuals seeking contact.)**

# The association of Genetic Support of Australasia Inc. (AGSA)

66 Albion Street
SURRY HILLS
New South Wales 2010
AUSTRALIA

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**Office Hours**
Mon - Wed: 9am - 5pm
Thur - Fri: 10am - 4pm



**Subscription Year 1st July - 30th June**

**ANNUAL SUBSCRIPTION**

| | |
|---|---|
| Individual | $24.00 incl. GST |
| Group/Organization | $48.00 incl. GST |

AGSA aims to:-

- provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group.
- facilitate access to individual support groups for those families with a particular genetic disorder
- provide a forum for the exchange of information between support groups regarding available community services
- educate the medical and allied health professional and the community about genetic disorders
- consult with government bodies, both Federal, and State, for appropriate funding for genetic services.

***The views expressed in this Newsletter are not necessarily those of AGSA***

If undeliverable please return to:
66 Albion Street, SURRY HILLS NSW 2010

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PP 242114/00005

THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

## CHANGE OF ADDRESS?

**PLEASE NOTE**

**To change your address on our record please fill in the new address below and return complete wrapper in an envelope to:**

**AGSA Inc.**
**66 Albion Street, SURRY HILLS NSW 2010**

**Name (Block Letters)** .....

**Address** .....

.....**State**..... **Postcode**.....