



VCFS & 22q11 Magazine

February 2010

President's Report

Hello all,

I hope you had a fabulous Christmas and New Year break. The committee has begun preparations for the 2010 Pink and Blue Cocktail Ball. It will be held at Cromer Golf Club (Near Dee Why) on the 22nd May 2010. We are in the process of collecting donations and prizes to be auctioned on the night. So if you know anyone or you yourself can donate a prize please contact Lucy Jackson. I would love to see as many of you as possible at the event. Keep a look out for details on our website coming soon.

2009 was a successful year for our foundation. We had the 1st VCFS walk for awareness, held Pink and Blue events in Awareness week and our TV advertisement was aired across Australia.

This year we are raising funds so that we can produce a booklet that will be published and sent to all Australian schools as well as given to parents to give to their children's teachers. The booklet will focus on education and social issues for those with VCFS. We also hope to have a section on the transition from school to work.

The awareness week will once again be held in the last week of August. We will send information out in June.

It was great to see family and friends come together for the Christmas party in December. A great day was had by all and the kids were thrilled when Santa arrived in a Fire Engine, thanks to the Willoughby Fire Brigade.

Last year the VCFS international conference was held in Rome and Dr Linda Campbell of Newcastle University has kindly written a report for us in this month's Magazine. The 2010 conference is to be held in Salt Lake City, Utah and I will be going in July. Dr Campbell is also beginning a new study on VCFS transition to adulthood. Details are in this magazine.

As you are all aware VCFS is a worldwide syndrome and recently Sherry Gomez author of "Missing Genetic Pieces" launched a new website for all those affected by VCFS <http://www.22qcentral.org/> this website is a useful tool and community directory promoting awareness and education of VCFS. The website came about after many years of planning and struggling as a parent of a VCFS child herself. It is a great site and I hope you all find it useful.

Speaking of websites, this year our foundation is getting an overhaul and Lucy Jackson in conjunction with Boyd design are currently reviewing our website and we hope to have a new look later in the year.

We value your input, so if you have any ideas or suggestions for the foundation please email me or one of our other committee members.

Maria

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Whats On

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VCFS & 22q11 Foundation Calender

- **General Meeting
Childrens Hospital
Westmead
14th March 2010 10am
RSVP Maria**
- **2010 Pink and Blue Ball
22nd May 2010
(See form on back page)**
- **VCFS Conference
22nd August 2010**

Become a Volunteer

Volunteering can be a rewarding job. The foundation can only continue to raise awareness with its volunteers. Contact the president at president@vcfsfa.org.au

VCFS & 22q11 Foundation Inc

Families & Professionals supporting those affected by

VCFS & deletion 22q11

Registered Charity CFN 13849

ABN 22 379 450 116

www.vcfsfa.org.au



Charlotte's Story

I just thought you might like to hear some good news about my (nearly) grown up daughter who has VCFS.

Charlotte is now 20 years old and started out with the usual problems of our children - palatial incompetence, Dyspraxia, learning difficulties (in her Primary school years she had Teachers Aide Special Support and she was in a special class in her high school years).

She also had scoliosis and when the bracing no longer worked she had to have rods inserted to support her spine and keep her straight.

However she was always kind, generous, artistic and had the sharpest sense of humour!!

She is now completing her 2nd year of a Floristry Traineeship where she travels daily by public transport and she has her L plates so hopefully she will attain her license.

Long ago the family took a video of her playing soccer with her pet rooster and she had asked me for years to send it to Australia's Funniest Home Video Show. She finally burnt it to a DVD, wrote down the address for me and said PLEASE post this!!

I did do that little task and earlier this month it was shown (a prize of \$550) on the show and it was voted the most popular clip and she won an amazing prize of \$10,000!! She is so thrilled about this and now is eligible for the semi final where the winnings are even greater, although she (in her own quiet way) is just happy that this wonderful win has happened.

I am so proud of her persistence - I am the usual busy, working mother who doesn't always do what she is asked straight away but Charlotte knew all along this video clip was really funny.

Parents with young children with VCFS might like to see that it's not such a bad diagnosis after all!!

Regards,

Mary Pearce

Do you like reading our family stories?

Supply is drying up fast. Please send us your story and photo so we can continue to publish them in our magazine.

Send your story to editor@vcfsfa.org.au

The foundation wishes to thank the following people and companies for their support

**Hatrick Catering
Wild Escapes
David Adler
Wine Brothers
Red Balloon
Chris & Margaret Hall
Krinklewood
Swissotel
The Beauty Tutor
Rose Only
Delorenzo Hair Care
Golf 4 U
Printa .net
David Sullivan
Audrey Currie
Creative Cakes by Julie
Emily Leiding Weight
Business computer Services
Louise Nade
Matt Nade
The Owl & the Pussycat
Childcare
Tribe Beauty Chatswood
Soft Play Chatswood
Dymocks Chatswood
Harmony Pharmacy
The Advertising Department
Bannisters & Rick Stein
Gordon Hart
Melinda Woods**

Chris got his Ls

..... now the fun really starts!

Well what a summer Chris Gunton (16) has had ... he volunteered for the Blind Cricket Carnival for two weeks, he did jobs like scoring, umpiring, and general assistance. He even managed to get on TV with Fox Sports Programme Inside Cricket, as they got the NSW cricketer Nathan Bracken to come out and wear special glasses to see if he could swing a bat without being able to see! We have a special interest in Blind Cricket as my husband Peter use to play when he was much younger, he isn't blind but he is visually impaired.

Then with lots of on line practise tests – and a few goes at the RTA – Finally Chris got his Ls!!! He is so proud of himself and we were all very happy! Another milestone we can tick off!

But now the fun really starts – driving! We did have a “go” in a car park with no cars, in the manual, but we didn't actually get to take off. So the next day we pleaded to the big brother Matt (18 ½) and asked if we could use his car (it's an automatic) – he finally agreed, and we went for our first ½ hour lesson. We managed to drive around the same car park for 5ks! So we have officially started the countdown of 120 hours. He has decided that he wants to learn on Automatic – which is fine by me, but it means we have update our car, it will take some time but hopefully we will get one real soon. I will be investigating into driving lessons too. One plus is that every 1 hour lesson you have with a driving school it counts as 3 hours of your 120 hours – that's a big plus eh? Will keep you posted on how we go!

Also this year is going to be a big one for Chris – he enters into Year 10 at Catholic Gilroy College Castle Hill. He will be doing lots of “transition courses” which is held over in Mt Druitt (hopefully we can drive to those); plus he will be doing work experience. He tells me has wants to do Green Keeping – I personally think we should look into something that involves Sport – what I have no idea – but if you have suggestions, you could email me at priscillagunton@optusnet.com.au

Thank you – cheers Priscilla, Peter & Chris Gunton

“Good things are not done in a hurry”- German Proverb

“The most useful virtue is patience”- John Dewey



Anxiety and Depression in Children and Adolescents with Learning Difficulties

For Parents, Teachers & Health Professionals

Dr Ann Wignall

Director North Shore and Ryde Child & Adolescent Mental Health and Clinical Psychologist
Tuesday, March 16, North Ryde

Venue: Stamford Grand, Cnr Epping & Herring Roads, North Ryde

Date: Tuesday March 16

Time: Registration 7pm, Presentation 7.30pm - 9.00pm (approximate)

Parking: Free onsite parking available

Dr Wignall's presentation will discuss anxiety and depression in children and adolescents, especially those with learning difficulties. Why are LD children and adolescents at such risk of anxiety and depression and what can parents, teachers and professionals do.

Cost: (Tea/Coffee included) \$ 20 LDC member \$ 40 non-member \$ 35 membership & seminar entry
GST FREE TAX INVOICE ABN 93 046 401 929

Please complete one form per registrant and retain a copy for your records
REGISTER ONLINE at www.ldc.org.au

VCFSEF 16th Annual International Conference Report

In July 2009, the VCFS Educational Foundation had their 16th Annual International Scientific meeting in Rome, Italy. The foundation, together with the hosting organisation Aidel22, had put together a fabulous program over three days. More than 50 experts, on VCFS from across the world came to present their research to a large audience consisting both of clinicians, researchers and many families affected by the syndrome. All presentations were simultaneously presented in English, Italian and French using professional interpreters - it was a fantastic experience. The VCFSEF, for those of you who do not know the organisation, is an international organisation that brings together professionals involved in the assessment, treatment and study of people with VCFS, families of those affected, and people with VCFS. The meeting includes both formal presentations, more informal sessions such as the 'lunch with the expert sessions' social opportunities and also plenty of opportunities for families to meet directly with experts.

A lot of research was presented from a variety of experts from many different fields such as counselling, immunology, neuropsychology, behaviour and genetics. Unfortunately, I cannot tell you all about it since I would have to write a book but luckily you can access most of the talks online on the foundations new website http://www.vcfsef.org/about_vcfs/powerpoint.html. I would, however, like to tell you about some of the presentations.

Professor Sarda from France described a group of 75 people of varying ages who all had a confirmed 22q11.2 deletion. He particularly pointed out that it is important to keep monitoring calcemia in adolescents and even adults with the syndrome since he noted hypocalcemia in 17% of the older people in the group. He suggested that hypocalcemia amongst more mature people with the syndrome is under diagnosed.

Dr Higgins from Upstate Medical University in Syracuse, USA described the protocols used at the VCFS International Center to care for children with VCFS. In particular she stressed that it is important to understand the natural history of the syndrome and to properly anticipate when different clinical features of the syndrome are expressed. If clinicians can do this it optimizes the care of the child and also minimises cost, pain, discomfort and risk. However, this approach requires a sound knowledge and understanding of VCFS from the clinicians involved in the care of the patients Professor Shprintzen drew the attention to growth curves for children with VCFS. He pointed out that although small stature has been reported as a clinical feature of VCFS this is not a common clinical finding. Rather, an atypical growth curve is often found. He suggested that children with VCFS grow at a different pace compared with children without the deletion. This is important to bear in mind since if children with VCFS fail to follow the growth curves developed for healthy children, inappropriate treatments with regards to caloric intake may be performed. Professor Shprintzen and Dr Higgins as developed a VCFS specific growth chart based on more than 1000 people with VCFS Dr Morrow introduced the International 22q11.2 consortium which is a large international collaboration for which blood samples from more than 1000 people with the syndrome will be collected. These samples will be used to try and identify genetic factors which influence the structure of the heart.

A study by a French group (Dr Campion) reported that severe hyperprolinemia may be linked with lower intellectual problems and the development of psychosis in people with VCFS. This has already been suggested by Dr Vorstman in Holland. Hyperprolinemia results in abnormally high levels of proline in the blood and is usually benign but has been associated with renal abnormalities, epilepsy and lower intellectual functioning. In people with VCFS this problem is associated with the lack of second copies of the PRODH and COMT genes... ..continued

Save the Date
VCFS Awareness Week 2010
23rd – 29th August 2010
VCFS Conference 2010

Conference Report Continued.....

Mr Debbane from Switzerland reported that social withdrawal as rated by parents and self reported increases in anxiety was associated with more psychiatric problems in adolescents. This study identifies important markers for clinicians to increase the possibility to predict the development of mental health problems. It also identifies potential treatment targets to prevent further unfolding of mental health problems. Furthermore, Professor Gothelf reported that people with VCFS and psychotic disorders had lower verbal intellectual functioning than non-psychotic patients.

Professor Swillen from Belgium reported that many children with VCFS have much more sleep problems compared with their siblings. In particular the children had more bedtime resistance, sleep anxiety, night waking, parasomnias and daytime sleepiness. She recommends that sleep problems in children with VCFS requires further research and clinical attention.

Finally, I should tell you about the study that we presented based on a study at the University of Newcastle of Newcastle that some of you have participated in. We have found that adolescents with VCFS have problems with basic social functions. In particular we have found that people with VCFS often don't use optimal strategies when looking at faces and therefore perform less well when they are trying to identify facial expressions. For instance, the young people in our study looked less at the eyes and more at the mouth compared to similarly aged peers without the deletion. This makes it more difficult to respond accurately in social situations and may make social situations more anxiety provoking than necessary. In other disorders where similar problems have been found, various therapies have successfully been trialled to teach a more appropriate way of looking at faces and similar therapies may be successful in VCFS although they have not yet been used.

It was a really good conference with a nice atmosphere and well-worth going to and I believe this year's conference in July which will be held in Salt Lake City, Utah, US will be similarly good.

Linda

Transition to adulthood: New study

This year we are launching a new study where we will be investigating how young people with VCFS are transitioning to adulthood. There have been many studies of children with the syndrome, primarily focussing on educational issues, cognition and behaviour and some on adults and in particular on the mental health problems that sometimes occur. These studies have greatly contributed to our understanding of the impact of a deletion at chromosome 22q11.2. One area that has not been looked at in depth so far is how young adults with the syndrome adapt to adult life.

Our study will look at how young adults with VCFS experience adult role functioning with regards to e.g., financial independence but also with regards to independent thought and free choices. We are also keen to find out their level of life satisfaction and coping skills that may be important factors in managing their physical and mental well-being.

The study will be carried out at the University of Newcastle, Australia this year but we are inviting you, regardless of where you live, to take part in the study! The study will be carried out using questionnaires and interviews via post or online, therefore distance is not an issue.

If you are interested contact us via post or email and we will keep you informed about the study.

I hope to hear from you,

Linda Campbell
Centre for Brain and Mental Health
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Newcastle, New South Wales 2300
Australia
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VCFS HAS NOT STOPPED US FROM DOING ANYTHING!!!

Transcript of a speech given at the 2008 VCFS Conference by Priscilla Gunton, a VCFS mother with a VCFS child

Part 2 – “My son, Christopher”

Christopher – Baby Hood

Now to Master Christopher – As a baby he was a very floppy baby, with no strength in his legs, he wouldn't stand up while you held him up and balance on your legs. He was very quite – loved sitting in his bouncer for long periods of time. He also loved laying in his stroller and never sitting up. He was delayed in all his miles stones ie sitting up at 9 months, walking at 22 months. I can't remember at what age he started to talk, but I'm sure it was late.

While Chris was in the non-walking mode I was advised to take him to the physiotherapist, she was the one that thought something wasn't quite right, she rang the paediatrician and told him so. Between the two of them it was decided that both Chris and I should be sent down to Genetics Clinic to be tested. The test came back positive to VCFS!

Finding Out

Seeing and meeting Dr Lipson I remember that appointment very clearly, I was asked to bring family photos along. I showed Dr Lipson all photos of my family and told him who did what. Basically he said since all of them went on to tertiary education then they wouldn't have VCFS. I remember feeling “Why me?” – and very cheesed off! I remember pointing out to Dr Lipson a photo of my brother Simon and saying what about him? As children we would be able to could be passed off as twins – but again he went on tertiary education and I didn't!

Feeling towards VCFS Since the diagnoses it has opened up many avenues of help/ support for Christopher, which I missed out on. Christopher has had support of the Early Intervention Program at Pendle Hill Primary School, Speech therapy, Funding through primary and high school, and later on transition programs into the workforce.

Health Worries

Facial Characteristics Chris and myself have the typical face of the VCFS – but there is nothing to be ashamed of there! We just look the same! Sometimes I forget and say to a teacher, oh I am Chris's mum – and they reply oh yes we know – the family resemblance is striking!

Asthma Cough Chris's asthma cough was very common during the winter months from 1 to approximately 12 yrs. We managed it well with preventative medication. We were only hospitalized once and that was because he got a virus on top of his asthma.

When he was about 8/9 he went through a couple of really bad winters, and we seemed to be down at the hospital, coughing our lungs out, finally our paediatrician told us that it was a “habit cough” – I didn't believe him at first, but when he talked to Chris about it, the cough finally stopped!!! So I think the paediatrician was right. However he still has asthma to a certain degree but it's not as bad as it used to be!

Behavioural difficulties Anxiety – Chris hates being late for anything especially the bus! He can't wait for his brother to leave school so he doesn't have to wait around for Matt!

Surprisingly no VPI Chris has not been diagnosed with VPI VPI after doing research for this speech I have discovered that Chris (a) can't blow balloons (b) doesn't have enough force to blow his nose properly (c) often mumbles and has been asked to repeat what he has just said – so am in the process of getting him re-assessed.

Delayed in gross motor skills Chris was behind other kids in his age group for milestones in running, riding a bike etc. His brother Matthew taught him how to ride a bike, after months of practise we thought he had it down pat! In Year 4 they had an excursion to a Road Safety place and they had to ride bikes! I deliberately didn't go on that excursion, but I told the teacher he could ride a bike, and had my heart in my mouth all day – pick up time came and Chris produced an “Encouragement Award” from the Constable! We were happy little vegemites! Then a couple of months later, while doing school canteen, with another mum, she and I started chatting and we got talking about that excursion and she said “Yes I remember Chris as I had to push him around on the bike!” – I went “oh dear!!!” – But since then he does ride bikes – doesn't like it – but will ride it occasionally – usually after a lot of pushing from his brother and friends!

We also attended group sessions at the Children's hospital to concentrate on his running/climbing abilities.

Delayed in fine motor skills Delayed in fine motor skills – he took forever to learn to tie his shoe laces, during Kindergarten a friend would drop her children off to my place on her way to work, and then I would take them on to school. Jack spent a whole term teaching Chris how to tie of shoe laces! He also had trouble doing up buttons on shirts too.

Phobias Chris has his fare share too – he hated the movies when he was younger usually because of the loud noise/dark and curtain; and he hates fireworks too due to the loud bangs!

School Life

Learning Difficulties He was diagnosed early ie 3 yrs old so that opened up a lot of windows!

Special Education Christopher started preschool at the age of 3 we had no separation issues. He was guided by a teacher's aide. We got into a programme called “Early Intervention Preschool” – 5 mornings a week – this was the year that he had turned 5. He actually could have gone to school that year, but it was decided to hold him back, especially because of the speech. The “Early Intervention Preschool” – was fabulous! He learnt so much! When the following's years intake came around, he was more than ready for big school! The Principal was surprised by the big difference!

Speech Speech therapy is very important – try and find a speech therapist that has had dealings with kids with VCFS. We continued to do speech therapy until the end of primary school. In the last couple of years they concentrated on comprehension and that assisted him greatly in the Basic Skills Test.

.....continued

During school We applied for funds for special education attention. We succeeded. He improved greatly at primary school – he finished Y6 with all C's in his report! He made lots of friends, represented the school in many sports – and generally had great time!

High School He is very happy at school – again has made new friends and have settled in well. He gets assistance with modified exams ie usually extra time, and if he wants he can have a writer/reader but he usually declines.

Just recently he has started Ice Skating for activities for sport. I was very hesitant with this choice because of the fact he has terrible balance. But his friends have been great, and have supported him throughout the sessions – just hope he doesn't break a bone!

Advantages for Chris being diagnosed early in life

Information Information on VCFS is easily accessible from doctors/and other health professionals also on the internet/books/published papers etc.

Education Chris receives help/support with speech therapy/early intervention/funding for school

Later in high school in Years 9/10 he will be able to attend transition courses which will hopefully lead to employment after school.

VCFS Support Groups Being part of a wider affected group who can be used as role models

Research Continues to the benefit of those affected.

Disadvantages for Chris being diagnosed early in life

Being different Just the realization that he has the syndrome and that he is different from the others.

Mum/Dad Mum/dad might be a bit overprotective for a bit too long.

Doctors Numerous doctor's appointments/repeated ENT infections especially when he was younger.

Future Possibly not becoming a father – but then again with the gains in research things might be a whole lot different in 10-15 years time.

Sport

Chris and Cricket Chris absolutely loves his sport especially cricket. His father Peter has a very keen interest in cricket and coached many years of cricket to both boys. Chris is very good being the opening batsman; he has a very good eye and hand co-ordination which allows him to catch some brilliant catches! He loves playing both outdoor and indoor cricket.

Chris and Soccer For many years Chris played Soccer. He could be very effective as a goal keeper... He loved the position of goalie because he didn't have to run the length of the field.

And his beloved Eels Chris loves watching football – especially his beloved Parramatta

Key people to support you with VCFS

Be the best advocate for your child Mothers knows what's best for their child

Paediatrician To keep an eye on all the health worries that is associated with VCFS

Special Education Teachers – Make them your best friends – still friends today with Chris's first special ed teachers from preschool to primary school. Bomb- bard them with books, web sites, any information on VCFS.

Speech therapy Very important – find one that knows about VCFS.

Gross Motor Groups Attend groups at the hospitals

Support Groups – so you know you're not alone!!!

Poets Corner



There was some confusion from readers about the “Mother From Hell” poem in the October 2009 issue of our magazine.

The poem was sent to a mother who was having difficulties with the principal at the school that her child attended. It was meant to be a cheeky way of cheering up the mother, by her child's psychiatrist, and to show that there is a way to bring about a change in attitudes by educating these narrow minded school officials. The mother was so impressed, she gave her doctor a framed copy of the poem for his waiting room wall.

“Progress is impossible without change; and those who cannot change their minds cannot change anything”

- George Bernard Shaw

Poets Corner

If you have any inspiring words you would like to contribute to the Poets Corner, please send them to editor@vcfsfa.org.au

What to do if your Toddler is not Talking Yet

by Lisa Baade



If all of your friends toddlers are babbling away and **your toddler is not talking yet** you may have reason to become concerned. One of the things parents often use to gauge their child's development is how early they start talking. Babies start off experimenting with noises from an early age, usually consisting of babbling noises, the typical "goo goo" and "gaa gaa" sounds, these will turn into "ma ma" and "da da". These babbling sounds eventually progress into Mommy and Daddy. So how do you determine whether your child has delayed speech development and what do you do if *your toddler is not talking yet*?

On a daily basis your child will find different ways to shape their mouth and change the noise they are making; the first step in their [speech development](#). Parents naturally spend a great deal of time talking to their baby's but they do this instinctually and mostly to enjoy the social interaction it offers. Many don't realize that this is also essential for the development of their speech and language skills.

Babies recognize the sound of their parent's voices from even before birth; they become familiar with us when they are in the womb. Parent's voices are comforting for them, especially for newborns before their vision is fully developed. Babies and toddlers mimic their parents; that's how they learn. The more verbal communication you engage in with your child, the faster they will learn.

By the time your child is the age of twelve to eighteen months your baby should be saying words like Mommy and Daddy. Over the next year of your child's speech development, they will learn to put together small sentences and communicate their needs to their family members. By the age of 21 months most children can be understood by their family. If your child cannot communicate with family members and be understood by the age of 30 months there would be cause for concern. At any stage, if your child is not meeting the age appropriate [developmental timelines](#) you should seek professional opinion. Trust your instincts, more often than not parents know best.

If **your toddler isn't talking yet** there are things you can do to encourage speech development. Keyword signing, otherwise known as [Baby Sign Language](#) has been shown by researchers to increase language acquisition by up to 3 months. Children using sign language have the language re-enforced to them by the parent as the gestures are used in conjunction with the word. Parent's who use sign language with their children are often more aware of their communication with their baby's and flood them with language on a daily basis. Gesture based baby sign language also [improves word comprehension](#) as the gesture is a natural movement that mimics the meaning of the word. Reading to your child is another important strategy that will go a long way to improving your child's language skills. When they are exposed to reading at an early age they pick up on the way language ebbs and flows and are more likely to have an interest in books later on.

So if you are concerned that **your toddler is not talking yet**, assess the developmental timeline as make sure they are reaching the recommended milestones. Not all babies develop at the same rate and the normal range can differ by months. If you are looking for ways to encourage speech development because your toddler is not talking yet, [keyword signing](#) and reading are both great places to start.

About the Author: Lisa Baade is the Author of Toddler Interpreter, A Parent's Guide to Baby Sign Language for Hearing Babies and Toddlers.

Teaching a simple keyword based communication tool such as [Baby Sign Language](#) can help to accelerate your child's speech comprehension and development. More importantly it can also provide an invaluable head start if you find out later that they suffer from a speech delay. For more information visit www.toddlerinterpreter.com

Lisa is very proud to associate herself and her company with our charity and will be donating 15% of all book sales sold through our website to the VCFS 22q11 Foundation. Thank you Lisa!



Q&A



Our son Tim is now 25 years old and recently been diagnosed with sleep apnoea. He has not been able to use a cpap machine and we have been advised that surgery may not give us any better results. Has anyone else been through this problem with sleep apnoea

Thanks. Shirley Adam

If you would like to submit a reply, please send it to president@vcfsfa.org.au

Suggestions will be published in the Parents Forum on our website www.vcfsfa.org.au





Around the Globe

"Philadelphia --- World renowned physician and paediatric endocrinologist, Dr. Angelo M.

DiGeorge, age 88, died Sunday, October 11, 2009"

Dr. DiGeorge first gained international recognition in the mid-1960's for his ground breaking discovery of the role of the thymus gland in human immunologic function. The congenital absence of the thymus and associated abnormalities is a birth defect widely known as DiGeorge Syndrome. The characteristics of the anomaly have led it to also be referred to as Velocardiofacial Syndrome, Shprintzen Syndrome or as the Chromosome 22q11.2 Deletion Syndrome since it was determined that most, but not all, of the affected individuals have a specific section of chromosome 22 deleted.

DiGeorge Syndrome, initially presumed rare, is now recognized to be one of the most common genetic disorders known, occurring in more than one in 4,000 live births. DiGeorge's original 1965 paper describing the anomaly has been cited by more than 500,000 medical researchers worldwide and a Google search yields more than 700,000 citations. During the course of his long career, DiGeorge was one of the key figures in transforming St. Christopher's Hospital for Children from a small community hospital into a nationally prominent medical institution. In addition to teaching thousands of doctors and medical students, Dr. DiGeorge's medical legacy includes his personal mentoring of more than 40 pediatric endocrinologists most of whom have gone on to achieve worldwide prominence in their own right. DiGeorge has authored more than 230 medical papers, abstracts and text book chapters and he has been an invited guest lecturer around the world. DiGeorge, a lifelong resident of Philadelphia, has received countless awards including being named in numerous publications as one of the United States' best doctors; and by Philadelphia Magazine as a World Class Philadelphian.

"We ourselves feel that what we are doing is just a drop in the ocean. But the ocean would be less because of that missing drop" – Mother Theresa



Medical Matters

"Dr Shprintzen, what is your opinion on issues faced by girls with VCFS?"

Your question is an excellent one, and certainly a multi-faceted one that has components of law, religion, moral codes, ethical considerations, and the inability to see into the future. First, keep in mind that there is not a one-size-fits-all approach here. I have a number of adult patients with VCFS who are in stable marriages with children, sometimes with VCFS, sometimes not, and in some cases, women who have undergone in vitro fertilization to assure a non-VCFS pregnancy. In some cases, these people have made wonderful parents and obviously being in a stable relationship has helped. In other cases, the worst case scenario problems that members of your group are worried about actually occur, and we see children placed for adoption, or in a dysfunctional home environment. If a person with VCFS has a severe cognitive or mental impairment when they reach adult life, then parents can seek permanent guardianship and then the medical decisions that need to be made are taken out of the hands of the patient. The more perplexing dilemma is what to do about the "typical" VCFS teen/young adult. Fortunately, permanent sterilization does not have to be a solution. There are effective techniques for birth control (as long as they are not forbidden on religious or moral grounds) that are essentially 100% effective, and may even have side benefits, such as mood stabilization. Birth control pills or implanted progesterones will provide contraception and squelch the severe mood swings that will often happen once a month. There is always the possibility that making a permanent move may be regretted later. Medical discoveries can be made that we are not thinking of right now, or someone can move into a stable, loving relationship that parents might not have expected, or they may prove to be better potential parents than anyone had ever thought. Bottom line, there is probably no right or wrong decision, especially when those decisions are guided by love, concern, and lots of thought about possible consequences (+ and -). It is worthy of a lot of discussion, and might be something important to discuss at upcoming VCFSEF meetings... Dr Robert Shprintzen



VCFS Family Christmas Picnic



I have always loved Christmas time. I love decorating the tree, having the house full of twinkling lights, the morning ritual of getting a chocolate out of the advent calendar, and of course the impending arrival of Santa. These days there is an extra date on the calendar that we look forward to each year. That is of course the VCFS Family Christmas Picnic, a time to meet up with our other family to share news and triumphs of the year gone by. It is some what comforting and reassuring to be able to speak to and share stories with other families who are travelling along the same road in life, and just understand!

We arrived at Willoughby Park, after only getting lost once. I immediately regretted wearing my flowing summer dress with the wind as it caught me as I stepped out of the car. This was our second picnic, and the kids immediately ran to the play area and were only to be seen when they needed food. Which I have to say was a great improvement on the previous year, where they hung to my legs the entire time.

After food was eaten, drinks were drunk, and many stories told, there was the arrival of one very special visitor. Much to the delight of the children, a red suit could be seen marching towards us. But alas, this was merely an impostor; the real Santa arrived in style with the help of the Willoughby Fire Brigade. What a treat, Santa and a fire truck! Santa joined the children to hand out presents to all those good girls and boys, and they must have all been very good this year as there were presents a plenty. The park was soon filled with many little fairies, cowboys and there were more rugby balls flying around than at Telstra Stadium. How does Santa do it?

I would like to take this opportunity to say a big thank you to Santa and all of his helpers for a great Christmas Picnic. I had two very excited and tired little girls, and we are very much looking forward to next year, and hope to see you all there.Audrey Currie

family

A D V O C A C Y

Moving Out.....from planning to action

A workshop about how you can support your family member with a developmental disability move into a home of their own with support

Who should come? Family members and friends of people with a developmental disability regardless of the level of support required. Topics of the workshop will include the planning process, housing options and exploring possible forms of support. NSW and Australian examples will be used.

Parramatta Monday 15 March; **Penrith** Tuesday 16 March; **Castle Hill** Wednesday 17 March ;
Gosford Thursday 18 March.

Registration 9am-9.30am, Workshop 9.30am-2.30pm, Cost \$15 per person. Morning tea, lunch and detailed resources are included.

Register by Thursday 4th March 2010

Phone for more information, venue details and to register on 02 9869 0866 or 1800 620 588 (freecall for NSW non metro callers) Email workshops@family-advocacy.com

Contact Us



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Ph: 9958 2578

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secretary@vcfsfa.org.au

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Editor: Chris Thorley

editor@vcfsfa.org.au

family

A D V O C A C Y

Hosts

in  **Control**[®]

What does Self Directed Planning, Funding and Support mean for people with high and complex needs?

A forum that unpacks what is required by government, services, people with disability and families to turn rhetoric into practice.

- 9.00 Registration
- 9.30 Robert Manga of People with Disability Australia, Judith Ellis of Ellis McRae and Margaret Ward of Mamre Association Brisbane, discuss individual, family and service perspectives on a self directed approach
- 10.45 Morning tea
- 11.15 A facilitated conversation on the challenges of implementing a self directed approach
- 12.30 Lunch
- 1.30 Self directed planning: a review of literature Trudy Van Dam of the Australian Catholic University
- 2.00 Options for self directed funding Karen Fisher of the Social Policy Research Centre
- 2.30 What does a self directed approach mean for government? Peter Shergold of the Centre for Social Impact, UNSW
- 3.30 Discussion
- 4.00 Close

Date: Monday 22 March 2010

Time: 9.30am - 4.00 pm (Registration at 9.00am)

Venue: Burwood RSL, 96 Shaftesbury Rd, Burwood NSW

RSVP: by 12 March

Cost: \$30 for professionals \$10 people with disability and families. (Inclusive of GST).

In Control Australia is a group of individuals and organisations who aim to bring about systemic policy change in Australia to enable individuals with disability and their families to manage their support. The organisation is an affiliate of In Control UK and works to provide an avenue for information exchange, critical inquiry, dialogue, collaboration, leadership and influence.

Please register and pay online at www.family-advocacy.com and click on the In-Control button on the home page.

Online reference code INC0310 will be required for payment

Minutes of VCFS Meeting Sunday 29 November 2009

Westmead Children's Hospital – Meeting Room

Attendees:

Maria Kamper, Louise Nade, Kim Clifton, Lucy Jackson, Chris Thorley, Mary Thorley, Slade Jenson, Pricilla Gunton

Apologies: Melinda Woods, Audrey Hickman Currie, Leanne Tye, Allis on Allo, Kim Clifton

Commenced 10.00am

Banking:

Membership email address to change to SJ

Paypal Direct to SJ

SJ to get a PO Box

Banking address to change to SJ

Slade to purchase banking software – QuickBooks Not For Profit

Sales of merchandise - MK to Check with MW re: BAS exemption & GST

Website:

Currently cost \$74.85 pa for domain name and webhosting

Photos to be 'saved for web' to ensure efficient downloads

Board happy with the new proposed layout for the website, which has been donated by Boyd Design, normal cost over \$3000. Content will remain the same.

Lucy to check that Boyd Design's donated website will continue at the same cost and level of accessibility

'Toddler Interpreter' has offered to donate 15% off all book sales to VCFS 22q11 Foundation. They will design a banner for the new website so they can monitor the amount of click thru's

Storage King logo – capitalize on their 30,000 hits per week

Links to VCFS22q11 on Twitter & Facebook to be on new website.

Magazine:

Due Feb – Mar 2010

Website update - Lucy

Xmas party Photos

Toddler Interpreter article – Lucy

Break up text with photos

Percy Puzzle Quotes

May Auction Event – Pink & Blue Fundraising Ball, Lisa Wilkinson attending.

Books for Libraries:

MW to discuss her thoughts, project delayed until next meeting

Web-linked access to books

Brochure online access to all libraries

2010 Aims:

School Starter booklet- Audrey Hickman to assist in the writing of.-Utilise CHERI resources

Grant – SJ & MK to write for assistance in financing new School Starter booklet

LDC – Allison Allo replaced by MK

AGSA (Assoc of Genetic Support of Australasia) - Mary Thorley check to see we are members

PR & Media – need to appoint one person to coordinate all PR & Media. Possibility of hiring a firm to handle this aspect of profile & fund raising. Pricilla to contact a possible PR contact.

Lucy to contact Today Tonight and Lisa Wilkinson.

Lucy to contact local paper Manly Daily for some local interest angle.

Maria to contact 60 minutes and follow up on last years possible story.

Leaving School/Education – additional information needs to be made available to members, experienced parents such as Pricilla, Leanne, Mary & Chris to assist is writing a contacts page. Nova Employment, Kumon and any other support opportunities.

Membership:

Review membership form - Add State - Include new PO Box address for SJ

2010 Fundraiser:

Date Sat 22 May 2010 Venue Cromer Golf Club - no room hire cost- "Pink & Blue Ball"

Lisa Wilkinson confirmed for an introduction

Need to arrange an MC -Live Auction - Silent Auction

Cash Bar but free drink on arrival

Bus along M2 \$5 per head, pre paid – MW to organise

Prize Call – Lucy Jackson to draft a letter requesting donations. Follow up from 2008's \$5000 donation from AstraZeneca.

Louise Nade to design a 'save the date' and 'invite' for the Pink & Blue Ball and use Lucy's personal PO Box address for the RSVP's.

Christmas Party:

13 Dec 2009 11.30am -\$10 per head per child

Storage King donated Benn Dunn footballs and hats

\$100 donated by Jessica Crawford

Beautiful Soles donated \$150 cash for Christmas gifts

Louise Nade to buy and wrap gifts

MK confirmed Fire Engine will bring Santa to the party!

Other Business:

None

Meeting closed 12pm

What's On



VCFS & 22q11 Foundation

General Meeting 14th March 2010 10am

Childrens Hospital Westmead

RSVP: Maria Kamper Ph: 9958 2578 president@vcfsfa.org.au

Pink & Blue Ball Saturday 22nd May 7.30pm

Cromer Golf Club, Cromer Rd, Cromer NSW 2099

Cost \$65 RSVP May 4th 2010 Lucy Jackson secretary@vcfsfa.org.au

PO Box 411 Freshwater NSW 2096 (see application form on back page)

Learning Links

February 22

Total Communication -This workshop will identifies specific strategies including use of visual aids and signing.

Time: 6.00pm – 8:30pm Venue: Learning Links, 12-14 Pindari Road, Peakhurst Registrations: Dana (02) 8568 8200

February 24

ADHD in the early years – understanding and responding- This workshop provides participants with an understanding of ADHD in young children **Time:** 6.00pm – 8:30pm **Venue:** Learning Links, 12-14 Pindari Road, Peakhurst **Registrations:** Dana (02) 8568 8200

February 24

Triple P (Positive Parenting Program)- Triple P is for parents who want to discipline less and enjoy their children more.

When: February 24; March, 3, 10, 17, 24 Time: Wednesdays 10.00am – 12.00pm Location: Learning Links, 12-14 Pindari Road, Peakhurst (Childcare available) Cost: Free Enquiries: Amanda Wheeler on 8525 8235

March 8

Working with families who are grieving - issues surrounding diagnosis of a disability in children

This workshop explores strategies for workers to use when supporting families who have a child with a new diagnosis

Time: 6.00pm – 9.00pm Venue: Learning Links, 12-14 Pindari Road, Peakhurst [Book online](#) for these discount prices:

Nonmembers \$131; Learning Links' Members \$121; Learning Links' Parent Members \$69 (all prices include GST)

Registrations: Dana (02) 8568 8200

May 5 and May12

Behaviour as communication; a functional approach - behaviour management for children with disabilities

This workshop aims to provide an understanding of the purpose of behaviour in order to teach and develop effective alternatives.

Time: 6.00pm – 9.00pm Venue: Learning Links, 12-14 Pindari Road, Peakhurst [Book online](#) for these discount prices:

Nonmembers \$131; Learning Links' Members \$121; Learning Links' Parent Members \$69 (all prices include GST)

Registrations: Dana (02) 8568 8200

Learning Difficulties Coalition (LDC)

Tuesday 16 March Anxiety and Depression in Children and Adolescents with Learning Difficulties

Registration 7pm Presentation 7.30 - 9pm (approx) Stamford Grand, Cnr Epping and Herring Roads, North Ryde Phone: 02 9806 9960 **Cost:**\$20 LDC member \$40 non-member \$35 membership (till June) and seminar entry info@ldc.org.au

Heart Kids

Family Camp 2010 -Registrations now open for next years Family Camp.

Mowbray Park **12th-14th March 2010** Free to members-Includes all meals and farm activities

Please RSVP to Dawn Everingham 0420 364 125 or dawn.everingham@heartkids.org.au

Regular Events:

Westmead Children's Hospital – Coffee Mornings

1st Tuesday and 3rd Thursday each month. Please contact Kim on 0406 420 627 or 9294 0800 for further details.

Sydney Children's Hospital Randwick – Coffee Morning Contact Karen on 0406 424 620 or 9294 0800.

CleftPals NSW

Morning teas If you are interested please email cleftpalsnsw@gmail.com

Family Advocacy

Moving Out From Planning to Action Workshops.

Parramatta March 15; Penrith March 16; Castle Hill March 17; Gosford March 18

Registration 9am-9.30am, Workshop 9.30am-2.30pm, Cost \$15 per person. Morning tea, lunch and detailed resources are included. Register by Thursday 4th March 2010

Ph:02 9869 0866. Email workshops@family-advocacy.com

Addults with ADHD (NSW) Inc

QUARTERLY AWARENESS AFTERNOONS for 2010 Saturdays 2.00pm to 4.30pm:

March 20 Venue: "The Muse", Sydney Inst. TAFE, Harris Street, Ultimo – Sydney

Gold coin donation at the door. Coffee & chat after speaker

SUPPORT FOR PARTNERS AND PARENTS OF ADULTS WITH ADHD

Quarterly Luncheons on the last Sunday – 12 noon-2pm

March 28, June 27, September 26, November 28

Venue: Macquarie Hospital 3/51 Wicks Rd Nth Ryde

Email or phone the office 02 9889 5977 / 0416 111 036

Playgroup Australia Inc

National Playgroup Week 21 March to 27 March 2010

To find out what's on in your state or territory during National Playgroup Week call 1800 171 882 (toll free)

www.playgroupaustralia.com.au



*Some of our VCFS Families
starting school and going
back to school in 2010!!*





THE 2010 VCFS & 22Q11 FOUNDATION

Pink & Blue Ball

SATURDAY 22ND MAY 7.30PM

CROMER GOLF CLUB
CROMER ROAD, CROMER
NSW 2099

\$65

DRESS IN PINK & BLUE
BALL GOWNS NOT REQUIRED

RSVP: MAY 4TH 2010

LUCY JACKSON SECRETARY@VCFSFA.ORG.AU
OR
PO BOX 411 FRESHWATER NSW 2096

Help us in "Making the Puzzle Easier"

I WOULD LIKE TO PURCHASE _____ TICKETS AT \$65 EACH
OR

I AM UNABLE TO ATTEND BUT WOULD LIKE TO DONATE
\$ _____ TO THE VCFS & 22Q11 FOUNDATION;

CHEQUE ENCLOSED FOR \$ _____

OR

PLEASE BILL MY VISA MASTERCARD

NAME ON CARD _____

CARD NUMBER _____

AMOUNT _____ EXP DATE _____

NAMES OF GUESTS ATTENDING

Thank You...

PLEASE DETACH AND RETURN TO...

SECRETARY
VCFS & 22q11 FOUNDATION
PO BOX 411
FRESHWATER 2096



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