fx info

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From the President

This is my first newsletter as President of the Fragile X Association of Australia. I would like to thank John Kelleher on behalf of the Association for his work as President over the past seven years. As you may know John has remained on the Board as Treasurer, Doug Rodgers as Secretary and Robyn Iredale has joined the Executive group.

I have been on the Board for three years and my link to Fragile X has been through John and Christine Kelleher and their family. My background has been in the financial markets in the UK, US and Australia.

I hope with the support of the Board and members to continue to increase awareness and understanding of Fragile X across Australia. I believe that the Association provides valuable support for people with Fragile X and their families through the children's and adult's clinics, counselling and information and support. Although this newsletter is not the appropriate place to list all the work of the Association it gives a sense of some of the many issues that we are dealing with on a daily basis.

Finally I would like to wish you all the best for Christmas and the New Year.

John O'Connor

Better Start - feedback

We have recently had discussions with Department of Families. Housing, Community Services and Indigenous Services (FaHCSIA) about Better Start. The Better Start for Children with Disability initiative provides funding for early intervention services for children who are fully affected by Fragile X. Children registered with Better Start can access up to $12,000 (maximum $6,000 per year) to pay for early intervention services including audiology, occupational therapy, orthoptics, physiotherapy, psychology and speech pathology.

We are interested in hearing about your experience of Better Start so that we can tell FaHCSIA if there are any problems in accessing the program. We are interested in any feedback including whether you been able to access the information you need to register, have you had any problems with the registration process, have you been able to find service providers in your area?

Please contact Linda on 1300 394 636 or email linda@fragilex.org.au with any comments.

Christmas/ New Year closing

The office will be closed from 4.00pm on Friday 22 December 2012 and will reopen on Monday 7 January 2013. For urgent calls during this time please contact the Telephone Support line on 0409 987 012.

Best wishes for Christmas and the New Year from the Board and staff of the Association.

Visit from Fragile X New Zealand

We had a visit in the office from Andrea Lee who is the National Coordinator for Fragile X New Zealand Trust. We were really interested to share ideas and hear about the clinical forums that they run in New Zealand.

Left to right Yasmina Lee , Andrea Lee, Joan MacDonald, Janie Roberts and Christine Kelleher.

The NZ Fragile X Clinical Forum is an opportunity for a discussion via video conference link between families affected by Fragile X Syndrome, paediatricians in their local area and Dr Andrew Marshall, Developmental Paediatrician and specialist in Fragile X.

Andrea had just been enthused and inspired by attending the Miami FX Conference and she shared some of her experiences with us.

It was great to meet Andrea and we agreed to continue our collaboration.

Global Gazette Disability and Ageing

The Global Gazette Disability & Ageing is published every two months and is distributed to over 19,000 people in the disability field in Australia and some key contacts in over 100 countries.

Board member Simone Zaia suggested that the Association approach the Gazette and ask them to include information about Fragile X as a way of increasing awareness in the medical and disability sector. We were delighted to be advised that the Fragile X Association will be the featured organisation in the Jan-Feb edition (Vol 7 No 37). The DVD will also be listed in the Education Resources section.

We are grateful to the Editor of the Gazette Ross Fear for recognising the importance of raising awareness of Fragile X. You can see the Gazette on line or subscribe by going to

http://www.globalcarestaff.com/page/global_gazette_disability__ageing.html

Melbourne families get together

Families in Melbourne recently organised a lunch as an opportunity to get together with other Fragile X families and share their experiences. The lunch was held on 21 October in Melbourne. The initial booking was for twenty people and there was some concern that not enough people would come but they were delighted when thirty people attended.

Parents and children came from Victoria, NSW and Queensland. It was a great lunch which every one enjoyed and they hope to organise another get together of families in 2013. If you would like to hear about future lunches please contact Bronwyn on bronwyn@seanwyn.net or the Association on 9977 0074.

Sydney Picnic

The Sydney Picnic was held on 2 December at Mona Vale. We were delighted that thirty five people came along on a fine, and fortunately not too hot summer day.

It was a great mix of families with some coming having their first opportunity to meet with other Fragile X families. Some really valuable links were made between families.

Rosie Donald takes on America

Rosie in LA

Janie was inspired by Rosie's stories about her experiences travelling by herself in America. Rosie has kindly agreed to tell her story.

What gave you the idea for the trip originally?

It all started when I first went to America back in 2002 for my brother's wedding. Ever since I wanted to go back and see more.

It was only 3 years ago that I decided I was definitely going to do it.

Where did you go?

I started in Hollywood, was there for 3 days, then to Pasadena for IMATS LA, then Anaheim for VidCon 2012, then Laguna Beach to chill out and Newport to also chill out!

How did you get to your destinations?

Plane, bus, car!

What were the best bits of your adventure?

VidCon was a huge YouTube event in Anaheim that was amazing!!

What were some of the challenges of travelling?

Getting lost and having to ask 100 people how to get somewhere, being homesick towards the end and money!

What would you like people to know about you and what you achieved?

You can do it! If you plan right, stay sharp and don't be afraid to make mistakes it'll be fine!

What did you learn about yourself and your capabilities?

I learnt that I'm stronger than I think I am and that I can get through my troubles one step at a time

What do you think of Fragile X and how it might have impacted on your life?

If I hadn't worked through my issues I'd be nowhere, period. Strive to be the best you can be!

Rosie in New York

Anything else that you would like to add?

Don't let something with a name define who you are or make you feel worthless. You are one of a kind there is no one else like you keep working to better yourself!

Janie Roberts—Counsellor

Counselling at Fragile X Association

Janie Roberts is the FXAA Counsellor and has a postgraduate diploma in counselling and psychotherapy. She is a clinical member of Counsellors and Psychotherapists Association of New South Wales Inc (CAPA) and Psychotherapy and Counselling Federation of Australia (PACFA).

Karen Loveday's award for service

On the 8 November 2012 Karen Loveday invited me to attend an award ceremony run by Disability Services Australia (DSA).

It was a very impressive event with hundreds of people attending and Mario Fenech and Matt Cowdrey giving inspirational speeches.

Karen was presented with a very special long service award for working at the DSA. She and her husband Neil had worked for the organisation for an amazing 30 years and their commitment and dedication was acknowledged.

Karen is part of the front line staff team and works on reception. Karen has said that she enjoys her job and has made some firm and long standing friendships while there.

Karen Loveday with her certificate at the awards ceremony

Karen's parents Pat and Will Van Der Mey also attended the ceremony. They told me how proud they were of their daughter who has created an independent life for herself while dealing with the challenges of Fragile X.

Telephone link up group

We are close to starting our first telephone link up group for women who are affected by Fragile X.

This is a really exciting opportunity for women, irrespective of where they are in Australia to have a chance to talk about their lives, share experiences and coping strategies and discuss issues that concern them. It will also be a chance to have a laugh together.

I will facilitate the groups and they will run for an hour once a week for 6 weeks at a time that suits everybody. If you or someone you know would like the opportunity to be involved please contact me on 1300 394 636 or janie@fragilex.org.au

I'd also love to have some telephone support groups for carriers, carers and siblings so if you are interested please contact me on 1300 394 636 or janie@fragilex.org.au.

Family to family

Since the last newsletter I have been able to link up more families who want to meet with or talk to other families in their area. If you are interested in contact with other families please ring me on 1300 394 636 or email janie@fragilex.org.au. I am in the office Monday, Tuesday and Thursday.

National Disability Coordination Officer Program

Over recent newsletters I have discussed some of the problems faced by people with Fragile X in finding employment. I thought I would highlight another resource which may be able to provide you with help in your local area.

The National Disability Coordination Officer (NDCO) program assists people with disability to access and participate in post secondary school education and training, and then subsequent employment, through a national network of officers, working within 31 regions across Australia.

"The NDCOs primarily work at the local level to reduce systemic barriers, build links and coordinate services between the education, training and employment sectors, so that people with disability have assistance at all levels and are subsequently able to better participate in further education, training and employment."

Some Regional NDCOs have produced resources, such as directories. A list of the current resources is available on the NDCO website under the 'Resources' tab.

You can find information about your local NDCO by going to http://www.innovation.gov.au/Skills/SkillsTrainingAndWorkforceDevelopment/NationalDisabilityCoordinationOfficerProgram/Pages/default.aspx or contacting the Dept of Industry, Innovation, Science , Research and Tertiary Education on 02 6213 6000

Linda Blair

In November Pearlyn, our Administrative Assistant left due to family reasons. I would like to thank Pearlyn for all the fantastic work she did, particularly in organising the papers for the audit.

I would like to thank our auditor Brian Tierney of Manser, Tierney and Johnson for his audit of our 2011/2012 accounts . The audited accounts are included with this newsletter for our members and are also available on our website.

Once again Pages and Pages are kindly stocking our Christmas cards and I would like to thank them for their continuing support. I would also like to thank our members and supporters who have already bought their cards. We have had such a run on the cards that we now only have the three trees design available. The order form is on page 12 of this newsletter.

Don't forget that if you have any news, stories or events for the newsletter please email them to me at linda@fragilex.org.au or contact me on 1300 394 636. You can submit your story at any time. The deadlines through the year are 15 November, 15 February, 15 May and 15 August but if something comes up after the deadline please contact me as I may still have some space available.

Liz Russell

I am really pleased to have joined the Fragile X Association as Administrative Assistant. I am looking forward to learning more about Fragile X and speaking to members.

I have varied experience in finance and administration, from small owner-operated businesses through to Deloitte and Touche of Eastern Europe.

I returned to the workforce in 2010 after raising three children.

I have also been involved with a number of volunteer groups as an adult literacy tutor and working to establish a Gardening Club at the local primary school. I am currently Secretary of my daughter's Local Scouts group.

Joan MacDonald

A Day at the Fragile X Association office

Its 8am Thursday morning, I work three days a week, Tues, Wed and Thursday. Janie works Thursdays too but I am in the office on my own this morning as she is off for a walk and talk counselling session with one of our members who is having a really hard time.

I check our voicemail then log on and answer emails. It's Linda's day off but she has emailed me this morning from her home address to remind me to write something for the newsletter.

I write up a report and actions from a meeting at Westmead hospital with Natalie Silove's team re the children's clinic there. I take a phone call from a woman who thinks she may have FX, I talk with her for half an hour then put together an FX info pack for her.

I open the mail and go through a children's clinic document pack from one of the families who are attending the next clinic. I see that there are several documents missing so I call them and explain what they need to do. A quick 10 minute lunch at my desk as usual and then I scan all the documents and disseminate them to the various therapists and doctors involved. Janie is back and we discuss my earlier call and what she can do to help.

I write a press release and get a press pack together then leave messages with some of my journalists contacts about some free PR for Mission to Lars which is being shown at the Brisbane Film Festival. I take a call from another children's clinic parent who wants to know where he can obtain a mental health plan that he needs to bring to the clinic. I put together a social story for one of the children who is attending the clinic, smiling at the photographs the family have sent me, such a good looking bunch our FX people! This story may help his anxiety levels and help him understand what is going to happen to him when he gets to the hospital.

I take a call from a journalist who asks me to send her my FX/Mission to Lars press pack and asks me lots of FX related questions, I tell her that this film is an amazing awareness vehicle for Fragile X and that is why we have tried to help them with a film distribution deal and getting the film PR in Australia. She wants to interview Kate (the producer of the film who is in the UK) and I give her Kate's contact details. I tell her I will Skype Kate that evening and set this up.
More emails to answer.

Its 4.30pm, time to leave the office, I haven't done half of the things I expected to do today but I am excited about the free PR for Fragile X and consider it a good day!

Mission to Lars update

The Brisbane International Film Festival featured Mission to Lars on 17 November. This was a great opportunity for Fragile X families in Queensland to see the film. It also continues to be a fantastic way to increase awareness of Fragile X. Stories about the film appeared in the Brisbane Times and thirteen local papers.

We are really pleased that there is so much support for the film. I keep in contact with Kate Spicer, one of the film makers so that we can be sure that we hear as soon as the film gets distribution in Australia. At this stage she hopes that this will happen in early 2013. As soon as I get more definite details I'll send information to all our members.

Resources

Companion Card

The Companion Card allows people with a severe and profound disability, who require attendant care at events and venues, to participate without having to pay for a second ticket for their attendant at affiliated organisations.

How does it work?
A Companion Card will be issued to people who are assessed as meeting all of the following eligibility criteria:

- you have a significant and permanent disability;
- you are unable to participate in most community-based activities without significant assistance with mobility, communication, self care, planning and the use of aids and other technologies does not meet those needs; and
- your need for this level of support is lifelong.

Cardholders present their card when purchasing tickets at participating businesses and are issued with a second ticket for their companion at no charge.

What is attendant care support?
Attendant care support includes significant assistance required with mobility, communication, self-care, or planning where the use of aids, equipments or alternative strategies do not enable the person to carry out these tasks independently.

The cardholder's chosen companion may be a paid or unpaid assistant or carer, friend, or family member or partner.

Can children apply?
Children may apply for the Companion Card however they must demonstrate a life-long need for attendant care support. As Companion Cards are given for life, no card can be approved before their life-long potential can be verified. As with all applications, children are assessed case by case and cards are not provided for supervision, social support or reassurance alone.

What organisations accept the Companion Card?
Affiliate organisations vary from State to State but include sporting events, cinemas, theatres, leisure activities, zoos and in some states public transport.

How do I apply?
To apply for a card you need to contact the State /Territory office where you live. The details are available at http://www.companioncard.gov.au/cardholders_apply.htm or by calling

Australian Capital Territory	(02) 6207 1086
New South Wales	1800 893 044
Northern Territory	1800 139 656
Queensland	13 QGOV (13 74 68)
South Australia	1800 667 110
Tasmania	1800 009 501
Victoria	1800 650 611
Western Australia	1800 617 337

Although you must apply for the card in the state where you live you can use the card in any State or Territory when on holidays or travelling.

Fragile X Family Experiences Research Project - Vel McKeachie

This is a preliminary notice to let you know that once I have my project approved at the University of Canberra, I will be inviting participants from across Australia to take part in a research project titled: 'Family experiences of delayed diagnosis of fragile X syndrome'. I am doing this research for a PhD at the University of Canberra, with the NATSEM research centre. Dr Laurie Brown is the chief investigator of the project.

My research is part of a larger project that is looking at the social and economic costs of fragile X syndrome. This interview guide will introduce you to the research aims, methods, time-frames, who will be involved, and an overview of some of the topics I hope to address.

Aims of the research

This is a qualitative study, so there won't be any testing or interventions. This is about gathering people's stories. I would like participants to tell or write their stories about their experiences of having a child with fragile X syndrome, before they knew the cause of their child's delays and other difficulties. The aim is to discover how parents (and other family members) compare 'not knowing' with how they felt and what their experiences were after 'knowing'; some families will have had multiple affected children so I will ask them to compare the different experiences with each child to see what changed, or how experiences might have differed, with an earlier diagnosis with a later born child.

My aim is to highlight the lived experiences, the ups and downs, the challenges and emotions of families from across Australia as they search for an answer. I hope that this information will provide detailed, rich examples of what it is really like to live with fragile X syndrome while not having a diagnosis, and the ramifications of 'not knowing'. This project aims to bring to life, through a holistic look at whole family situations, statistics and other quantitative data that have been presented in previous research papers about families living with fragile X syndrome.

Methods

My research methods will involve a short pre-interview questionnaire to gain demographic data about the participants, including the family constellation and number of other known fragile X affected extended family members, as well as 'costs'. I will then conduct in- depth interviews with people who volunteer to take part, either in your home, or somewhere that you nominate and will feel comfortable having a conversation with me and telling your family story. The interviews will take 1-2 hours, and there may be follow up interviews in person or over the telephone to clarify any uncertainties that may arise, or to expand on some topics if you feel able to do so.

I am also interested in having participants who feel inclined to write their stories. This may appeal to people who would like to spend time contemplating and recalling, without feeling pressure of the time restrictions necessitated by an in-person interview style. Written stories can be developed as ongoing journals as you recall and discuss with family members some of the events and experiences and impacts in your lives of 'not knowing'. The time frame for finishing stories will be a few months during the data collection stage of my research in 2013. The research will take place over the first half of 2013, with possible follow up in the latter part of the year. Results will be written up in 2014. A summary of results may be requested by participants.

I am interested in looking at whole families to really gain an in-depth picture of their unique situation, so am inviting both parents, and if appropriate, children in the family including fragile X affected children, non-affected children and siblings. Grandparents and other extended family members who are affected are also invited to participate. Parents will be asked to sign a consent form for themselves and, if appropriate, to allow their children to participate (only if they want them to, and if they are able). Parents will be asked to guide the researcher towards appropriate involvement and limitations with their children to enable me to work sensitively with their needs and 'comfort zones'. I won't be doing any 'experiments' or interventions, but simply asking about their experiences of not knowing about fragile X syndrome as a cause of symptoms and what that felt like.

I would like to include both fragile X affected boys and girls, and where possible, families with siblings who were diagnosed much earlier than the first child to compare the experiences of delayed diagnosis with early diagnosis. I am interested in including people with a range of cultural and social backgrounds, a range of age groups, and a range of degrees of fragile X. I hope to travel to rural, outback, urban and other places to meet with families.

If you are interested in taking part in this project, or would like more information about it, please email, phone or write to me:

Vel.McKeachie@canberra.edu.au
(02) 6201-2014 (leave message with your phone contact)
Vel McKeachie, c/o NATSEM
University of Canberra,
Bruce, ACT

I look forward to hearing from you and meeting people who would enjoy working with me in a relaxed, informal and conversational type of interview where your stories and experiences are considered to be important, and where parents are considered to be the 'experts' about their family situation.

Trials and studies

Clinical Trials

There are currently ongoing studies for both adults and adolescents with Fragile X at the following centres. Contact your nearest centre if you would like further details

Study for adult participants (over 18 years old)

NSW
Hunter Genetics, Waratah NSW
Contact: Jackie Boyle & Carolyn Rogers (02) 4985 3136

Royal Rehabilitation Centre, Ryde NSW
Contact: Jackie Boyle & Carolyn Rogers (02) 4985 3136

Victoria
Fragile X Alliance, Caulfield VIC
Contact: Melanie Van Buuren (03) 9528 1910

Study for adolescent participants (Between 12 & 17 years old)

NSW
Child Development Unit, Westmead Children's Hospital, Westmead NSW
Contact: Dr Natalie Silove on (02) 9845 2829

Victoria
Murdoch Childrens Research Institute, Parkville VIC
Contact: Dr 'Eppie Yiu' (03) 8341 6374

Social Research Study

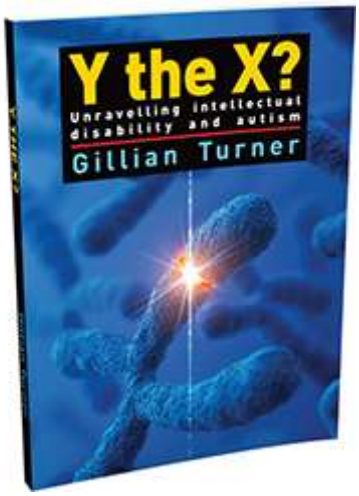
An Australian Study of families who carry the fragile X gene

The Australian Research Council (ARC) has funded a 3 year study to help families, health professionals and educators understand how being a carrier of fragile X impacts on Australian families. If you are interested in this study please contact the study team in Melbourne or Sydney

Claudine Kraan (Melbourne)
Monash University
School of Psychology and Psychiatry
(03) 9902 4162
claudine.kraan@monash.edu

Rachael Birch (Sydney)
The University of New South Wales
Department of Developmental Disability Neuropsychiatry
(02) 9931 9160
fxtas@unsw.edu.au

Y the X? Unravelling intellectual disability and autism



This book by Gillian Turner, Emeritus Prof of Medical Genetics Newcastle NSW, MD, FRCPC, MRCPE, B.Sc, AO reveals the approach by which the major causes of intellectual handicap have been discovered over the last fifty years.

Gillian Turner is an Australian geneticist who was involved in many of the important discoveries both nationally and internationally. Gillian has had a long involvement with Fragile X and the FXAA is grateful for her work in having Fragile X identified in Australia and establishing the GOLD Service in Newcastle.

Gillian became interested in X-linked intellectual disability or XLMR in the late 1960s when working at the assessment . This book follows the path that she and others took to identify Fragile X and other genes on the X chromosome that are involved with brain function.

It outlines:

- The process by which abnormalities of small and smaller parts of chromosomes are identified.
- The work of finding the amazing number of genes on the X chromosome involved in brain function.
- The long journey unravelling Fragile X.
- The importance of new point mutations originating from older fathers.
- Speculations about the causes of autism and how the male preponderance and increasing prevalence can be explained.
- The importance of the X chromosome in the evolution of homo sapiens.

After reading this book you will have a grasp of the basics of genetics and understand the excitement of research and the part so often played by chance.

The book is available from ythex.mybook.net.au RRP $30 + P&H. There is also a copy in the Association's library which is available for members.

Calls for Fragile X to be included in newborn screening

Researchers from Murdoch Childrens Research Institute say Fragile X syndrome could be the first genetic disorder in the last decade to be added to newborn screening. This is thanks to an early detection test which the researchers have shown works on archival blood spots collected from newborns that later developed Fragile X.

Newborn screening involves taking a blood sample from the baby two to three days after birth and testing for a range of conditions that will benefit from early diagnosis. The last time a new test was added to newborn screening, was in 2002. Newborn screening currently tests for 25 conditions.

The world-first Fragile X test, which analyses a DNA region that was previously thought to have no function, can both diagnose Fragile X syndrome and predict the severity of symptoms. Prior to this test, it has been difficult for doctors to diagnose Fragile X until the age of three years or older, particularly in girls. Importantly, early identification and intervention should increase the potential of children with Fragile X syndrome.

In the study, which was published in *Genetics in Medicine*, researchers used the early detection test on fresh blood and archival blood spots from 588 adults, children and newborns, including 242 with the Fragile X full mutation, and found the test was approaching 100% accuracy in both males and females.

Lead researcher, Dr David Godler said the test was highly advantageous for newborn, infant or early childhood population screening because the test detects those who would directly benefit from early diagnosis.

“The case for Fragile X to be included in newborn screening has been strengthened by evidence that early intervention with psychological and education therapies, and treatment with drugs can improve symptoms in affected children,” David said.

“The other important advance has been the development of a suitable test. Full consideration of a Fragile X heel prick test in newborns is now unconstrained by the lack of a suitable screening tool for this purpose. The comprehensive cost–benefit analysis of Fragile X testing within newborn screening programs can now be fully addressed.”

Researchers estimate the cost for inclusion of Fragile X syndrome would be between $700,000 to $1.5 million for every 100,000 babies tested. Based on the frequency of the condition, between 21 and 38 affected individuals would be identified in this population.

The societal cost of providing healthcare services over the lifetime of one individual with Fragile X syndrome is estimated to be between $350,000 and $2.5 million. Its inclusion into the newborn screening program has the potential to reduce the incidence of Fragile X in the population through identification of at-risk families who would be provided with reproductive options, and could see a very significant reduction in healthcare costs for the condition.

Fragile X can be compared with Cystic Fibrosis, another genetic condition which is currently tested in newborn screening and identifies 25 affected individuals per 100,000 babies. Screening for CF aims to achieve an early diagnosis and earlier treatment of symptoms. It also benefits these children’s parents through being informed of reproductive options.

The estimated cost per individual over their lifetime is $350,000. Current testing for cystic fibrosis costs about $700,000 for every 100,000 babies screened.

“Given these similarities, the same arguments can be put forward to support newborn screening for Fragile X,” David said.

Fragile X Articles

Dr Jonathan Cohen, Dr Alison D Archibald, and A/Prof Sylvia A Metcalfe PhD, BSc (Hons), have published two articles on Fragile X which appear in the Medical Observer.

The first deals with clinical presentation and diagnosis and can be found at

http://www.medicalobserver.com.au/news/fragile-x-syndrome-part-1

The second article focuses on the assessment and management of fragile X

http://www.medicalobserver.com.au/news/fragile-x-syndrome-part-2

Christmas Cards

1. Xmas Trees (12.5 cm x 12.5 cm)

2. Presents (11.6 cm x 17 cm)

3. Angels (11.6 cm x 11.6 cm)

$8.00 for a pack of 10 cards.
Message inside the cards:

I would like to purchase _____packs @ $8 each = $_____

+ Postage $_____$2 per pack (with a max. postage charge of $10)

TOTAL COST = $_____

No. of packs: _____

Please make cheques payable to the **Fragile X Association of Australia**
or please debit my MasterCard / Visa (Please Circle)

Expiry Date ___ / ___ Name on Card _____Signature _____

Please fill in your details:

Name:_____ Ad-

dress:_____

Suburb: _____Postcode: _____Telephone:_____

Please mail to Fragile X Association, PO Box 109, MANLY, NSW 1655 or fax 02 9976 2396