# Fragile X News

No 11/05
SEPTEMBER 2005
Fragile X NEWSLETTER

Hi everyone,

This newsletter is put together with the assistance of the Fragile X Association of Victoria (parent support group), the Fragile X Alliance Inc. and Genetic Health Services Victoria. We hope you enjoy the newsletter.

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## GENETIC HEALTH SERVICES VICTORIA

### Samantha Wake

As a genetic counsellor with Genetic Health my role includes facilitating genetic counselling and genetic testing for families where there is a diagnosis of Fragile X, and providing support and information about Fragile X Syndrome. Please feel free to contact me (I usually work Wednesday to Friday) at any time if you have any queries.

FXAV Fragile X Association of Victoria

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Dear Friends,

**Welcome** to the Spring edition of our newsletter, such a cheery time of the year with flowers going into bloom and the promise of warmer weather ahead, I love it!

The present team that runs the FXAV have been working together since early 2003 when we took over the reins from Mary Lloyd who had been running the organisation almost single-handedly prior to then and did a commendable job under the circumstances! Just to highlight what our group is all about, the main focus of our team is to reach out to you and assist by way of organising seminars, articles in the newsletter

on specific FX related issues , and organising events whereby FX families can meet others in similar circumstances, and share and feel less isolated and alone in their predicament eg coffee morning group, family fun day and of course the peer counselling service which is run by a group of parents with FX children and adults.

All families needs and issues can be quite different, and we are interested in hearing from you if there is a particular area you would like addressed either in the form of a seminar or newsletter article etc. We endeavour to cover topics that we feel will be of value but feedback from you the families is most important to keep us on the right path.

Another huge goal of the FXAV is to raise the profile of FXS and make it as recognised as Down's Syndrome and other well known conditions like Cystic Fibrosis. Because the occurrence of FX is almost as common as Down's Syndrome we find it difficult to understand why it is still so 'hidden' and unheard of in the community and are looking at ways of addressing this. In the past there have been a couple of articles printed in 2 major newspapers and 2 major magazines centred on a couple of families personal stories of living with FX (my family included) coupled with the medical perspective given by Dr Jonathan Cohen. Currently we are trying to raise funds so we can design and print off posters and brochures about FX and distribute them to all the doctors and paediatrician's waiting rooms in Victoria. We are also waiting to receive a batch of FX brochures from the Fragile X Association of Australia which is based in NSW to be distributed to all the Specialist Schools in Victoria to make Special Ed teachers more aware of the condition.

We are always looking for ways to raise funds for projects like these and if anyone would like to help with ideas or indeed by holding a fundraising event for us ( with our help if needed) eg a charity fun run (or walk as in my case!) we would be extremely appreciative and eager to assist. Arnold and Jeannie Pacifico who are FXAV members put on an annual Xmas lights display at their home in Bellevue Boulevard in Hillside which is incredibly spectacular, and raise money for our group in that way for which we are very thankful.

Recently Marcia Braden, a psychologist from the U.S., with a specific interest in FX, presented an evening seminar dealing with issues surrounding teenagers and adults with FX. It was an interesting and informative night and equipped parents and carers with useful insights, knowledge and advice they could take home and implement.

I always look forward to the coffee morning group and last month it was lovely to meet a couple of new faces, both mums having a son with FX, and to hear their stories and share with each other. We meet about every 2 months so please contact us if you would like to come along and meet other parents.

## Up coming events

**FXAV AGM Oct 2nd, 2005**
The FXAV Annual General meeting will be held on October 2nd, 2005. This meeting is open to any interested individuals so please contact me if you would like to attend. **For further details** see enclosed flyer.

**Evening Seminar/Discussion Forum with Louise Gane**
**Thursday 10th November**
Louise Gane is a renowned Genetic Counsellor from the U.S. with a rich and broad knowledge of FX gained from studies, research and working with many FX families. This promises to be a very interesting night and Louise will be most happy to advise on any questions you may have relating to FXS.
**For more information** see enclosed flyer.

**Family Fun Day**
We have decided to change the time of year when we hold the 'Family Fun Day' to March/April due to the unstable weather in November. If anyone has ideas on venues for this event we look forward to hearing from you. Ideally the venue would be 'childsafe' ie protected from roads by an enclosure and away from open water. Also BBQ facilities would be a requirement. We will send out details for our next Family Fun Day when all the arrangements have been finalised. Thankyou.

**FXAV Webpage**
For current information and notice of events please log onto
www.fragilex.com.au/fxav

As some of you may know, I have made the decision to stand down as President of the FXAV at our next AGM. I will continue to serve on the committee as a general member and look forward to helping build upon our past achievements and being part of a progressive and warm and friendly group. I would recommend to anyone to join in the work we do. For me it has given me a greater sense of confidence and belief in myself, experience in organising skills, leadership skills as well as the satisfaction of knowing I have helped to make a difference. My brother in law Andrew, who is the Treasurer of the FXAV, has written an article for this newsletter too about the benefits of volunteer work; he is very committed and contributes greatly to our group and enjoys being able to give back to the community.

On that note I would like to acknowledge the FXAV team and offer a HUGE THANKS for their continued support, ideas, efforts and commitment, thanks go to Sharon Davey, Andrew Bartle, Joy Burkinshaw, George Tziotis, Lindsay Coates, Arnold & Jeannie Pacifico and Dr Jonathan & Rashelle Cohen. I would also like to extend warm wishes and thanks for past efforts to John & Gwen Waters who have been unable to attend our group recently due to ill health. We all wish them well for the future. Thanks also go to Alison Dreckman for her assistance in organising the coffee morning group.

This will be our last newsletter for this year so until next time take care.

Best Wishes

Maggie Bartle
President, FXAV

# THE FRAGILE X ASSOCIATION OF VICTORIA NEEDS YOUR HELP!!

## Andrew Bartle

The Fragile X Association of Victoria of which I am a proud member, aims to unite the Fragile X community within Victoria to enrich lives through educational and emotional support, promote public and professional awareness, and the support of advanced research toward improved treatments for Fragile X Syndrome.

This committee is staffed by members of many families either directly or indirectly affected by this syndrome, each of these families are investing their own time participating in the search for supporters, benefactors and philanthropic donations from public and privates sources, as well as following up fund raising initiatives of our own all aimed towards funding our awareness activates.

**We aim to …**

- Provide support and information for Fragile X families from those who share and understand their concerns and needs,
- Educate, inform and advise the public and professional people about the prevalence and nature of Fragile X in order to raise awareness and understanding of the syndrome and improve the care of all people affected by Fragile X,
- Support research into all aspects of the syndrome and assist in the publicising of the results.

**We do this by …**

- Providing local contacts for Fragile X families,
- Supplying comprehensive information about Fragile X without charge to Fragile X families and support peers,
- Providing a national peer counselling helpline for education, family welfare benefit guidance and adult services,
- Hold and or support information conferences and briefing sessions,
- Publish booklets and papers, reports of conferences and a regular newsletter,
- Creating awareness of Fragile X through media campaigns and distribution of literature,
- Promote Fragile X families involvement in research and publishing the results of these studies,
- Encouraging the transmission of research findings from abroad.

The reality is that we are only a small team and our effectiveness as group is very much limited by the size of our membership.

As a team we all got involved with the Association as individuals for different reasons, some of us are parents of children affected by

Fragile X and have experienced first hand the emotional pain and frustration that such an affliction imposes on Fragile X Families. The FXAV needs more members in all aspects of our operation, not just on our committee, and we need more volunteers. A volunteer does not have to be a parent or even a family member of a Fragile X affected person. A person can volunteer for many reasons.

Volunteering can have a meaningful, positive impact on the Fragile X community as a whole. But do you know that it can have many benefits for you, too?

You can gain valuable life experiences and skills. Whether you become a member of the FXAV committee or help out on fundrasing or seminar activities, you'll experience the real world through hands-on work. You can use this experience to explore personnel or career interests. You will meet interesting people as we hope the FXAV programs and activities will bring together a variety of people from all walks of life.

Both the recipients of your volunteer efforts and your co-workers can be rich sources of insight. Volunteering has many other intangible benefits. It can help you give back to society, break down barriers of misunderstanding or fear, explore personal issues, and even have fun.

Volunteers are the heart and soul of the FXAV. Through their efforts, the FXAV can effectively staff and support education programs about Fragile X for patients, families and the public, and increases awareness of the Association and Fragile X in general.

Join our current team of volunteers who use their leadership, communication and organisational skills to run successful activity programs and fund raising events. The leadership team (President, Vice President, Treasurer, Secretary and Committee Membership) work together to oversee fundraising, education and awareness programs.

Join our team of volunteers and help coordinate seminars, information/awareness events and raise funds for Fragile X research and support needs. Our Committee develops and leads fundraising projects.

Join our team of volunteers to help to increase awareness and educate individuals with Fragile X, their families, health professionals and the general public about the FXAV and Fragile X.

CONVINCED!!!! EXCITED!!!

If you are interested in helping us at the FXAV please contact us.

## Internationally Renowned FXS Genetic Counsellor

## Louise Gane to visit Melbourne

Louise Gane, who is internationally recognised as one of the world's leading figure in genetic counselling in FXS, will be visiting Melbourne this November as a specially invited guest of the Fragile X Alliance Inc. She will be giving a special presentation to families on the evening of Thursday 10th November

entitled “Genetic Issues for Families with FXS” in which she will discuss many of the problems and uncertainties, as well as the joys, that we all face having FXS or a family member with FXS.

Since 1984, Louise Gane has worked with families who have a child or children diagnosed with fragile X syndrome. In 1991, she joined the team led by Dr Randi Hagerman at the Fragile X Treatment & Research Centre in Denver, Colorado as a genetic counsellor. Under the directorship of Dr Randi Hagerman, Louise is now located at the UC Davis M.I.N.D. Institute and is involved in counselling patients and their family members for fragile X syndrome and the newly described FXTAS (Fragile X Tremor Ataxia Syndrome): helping them to deal with and understand the diagnoses, assessing genetic risk, explaining reproductive options, reviewing the latest research findings and directions, and addressing psychosocial issues related to both diagnoses. She is also Development Officer for the M.I.N.D. Institute raising funds for ongoing research into fragile X syndrome and FXTAS. She lectures nationally and internationally and is co-author of many clinical and scientific publications related to fragile X syndrome and genetic counselling.

Louise has more experience with families than probably anyone else known and has seen well over 1,000 families, and stays in close contact with virtually all of them. Topics to be covered in her visit will include pedigree analysis, questions to ask patients re family members, issues associated with each family member, testing priorities and methods, and research at the MIND as it relates to the families.

There will also be opportunity to ask any question you may have about yourself or your family. We strongly encourage you to come along to this event, in order to take the opportunity to benefit from Louise’s unique and very personable style.

(NB Please use the booking form included with this newsletter to ensure you don’t miss out.)

***Tea & Coffee Mornings***

A small group of us have started getting together every month for a tea & coffee morning. Nothing official about it, just a good excuse to spend some time with people who also have children with Fragile X. If you’re interested in joining us, please call Alison on: 9569 3603.

# The Fragile X Association of Victoria Peer Support Network

**The Peer Support Network welcomes you to our service, so please don't hesitate to contact us to share any issues or difficulties you may be experiencing.**

| Peer support member | Fragile X children/ adults | Other children |
|---|---|---|
| Elizabeth Tziotis Temple-stowe | Guardian of grand-daughter 5yrs | 2 adult daughters, one adult son |
| Marie Pullen Rowville | Son 9yrs | Daughter 6yrs |
| Joy Burkinshaw Mornington | Adult son | Adult daughter |
| Maggie Bartle Frankston | Daughter 18 yrs. Sons 15yrs & 4 yrs | Son 15yrs |

**Contact details for the FXAV Peer Support Network**
**Tel: 0421 152 690**
Please call this number and leave a brief message stating with whom you would like to speak and we will return your call a.s.a.p. Thank you.

# Siblings & Understanding Disability

## Mary Lloyd

"Before I learned about autism, it was confusing and hard to understand why my brother seemed different from me and from most other kids I knew. I didn't really know why he talked out loud to himself, or copied what I said and repeated it right back to me, or flapped his hands in the air when he got excited."

It is very common for siblings to feel unsure about their brother or sister's disability. A lack of understanding can leave children feeling scared, embarrassed, guilty and confused. Taking the time to explain the disability in language that is easy for them to understand will help to offset these feelings. Explaining disability or illness can be difficult. You need to take into account the child's stage of development and their ability to comprehend the information. Take the time to plan your explanation and use simple language.

It is important to explain disability to children of all ages. Very young children are easily confused and they can make the wrong connections between things or events. For example young children will often mistakenly believe that they can catch a disability. The fear of something being contagious is common for young children. You only have to think of the number of times you've heard a young child say something like, "I'm not touching that, that's got Girl germs!"

Very young children also have a tendency to link things or events to themselves. For example when a young child is told that their parents are not going to live together any more, it is common to hear a child say something like, "Please mum I promise I'll be good, I won't be naughty any more, I promise!" In this case the young child has linked the fact that the parents are separating to their behaviour. In the case of siblings who have a brother

or sister with a disability, it is not uncommon for a young child to think that they may have done something to cause their sibling's disability.

It is often difficult for adults to imagine how children can make these connections. You can find many examples of these errors in the sibling literature. In the following example a young girl believes that playing with her hat has caused her brother's epilepsy.

"He was playing with my hat and then went very quiet under the dinner table. There was such a panic in the house, it was Christmas lunch. I never let my brother play with anything of mine after that — it was an obsession — in case it caused another seizure."

When children reach primary school they begin to notice differences between each other. It can often be confusing when other children make comments or ask questions about their sibling's disability. These questions and comments may be the first time that siblings begin to understand that there is something different about their brother or sister, and that other kids don't have siblings with the same differences like theirs.

"I had grown to understand that Em was 'special', but it wasn't until I was in primary school that I began to notice real differences between Emily and myself. Comments such as "Your sister's a retard", "spastic" and "mental" became common to me around the school yard. Finally, the penny dropped. My sister's disability wasn't just something that meant she couldn't learn as fast, didn't always behave as well as other people her age, and had to wear glasses. In the school yard it wasn't looked on as such a simple matter. This was a bigger deal than I thought."

Questions about a sibling's difference aren't always meant to be unkind. Children learn about their environment by asking questions. Having a good understanding of their sibling's disability, and being prepared with a simple explanation can be very helpful when other children ask those unexpected questions.

For example when my daughter was in prep her friend asked her why her brother jumps up and down and flaps his hands. She responded "that's 'cause he's got Fragile X." My daughter's response at age five was simple and reflected her level of understanding of her brother's unusual behaviour. The problem arose when her friend had a follow-up question, "What's that?" To which she answered "Um, I think it's a disability thing". Not satisfied with this they decided to ask her friend's older brother who was in my son's class. He answered their questions by saying "he's got brain damage". The result was one confused little girl who didn't really know what to make of this information, except that it didn't sound good.

Giving children simple explanations using language that is familiar to them can help to offset feelings of confusion or embarrassment. Examples of this may be:

"He's got a disability. It's called Fragile X. It means that he does different stuff like flapping his hands when he gets excited, or when he's happy. He's a really good reader! He reads the TV guide every week. It's cool."

"She's got cerebral palsy. It means her legs don't work properly. She

can't stand up so she has to use a wheel chair. Sometimes mum lets me ride in the chair. She can't speak, but she can tell me stuff anyway."

"He's got autism. He does different things to me. He can talk a bit. He's learning slowly. He says words over and over. Sometimes he gets mad because he doesn't understand. Like if I play with his stuff he doesn't like it. But he loves computers and he can climb better than any kid ever."

"It's called epilepsy. It means that sometimes she has fits. That's when she falls down and her body shakes a bit. Sometimes she just stops and stares for a little while. When she has a fit she can feel really tired and she needs to have a sleep. But when she wakes up she's okay."

Finding the right words to explain a child's disability can be a challenge for parents. The trick is to keep it simple, but to give enough information to explain the differences whilst trying to throw in some positives as well. As children get older they may ask more questions in order to get a better understanding. Just keep giving them little bits of information and check if they understand you.

When children enter the teenage years they tend to ask questions that help them understand what the future might look like for their sibling with the disability. At this age they are trying to build a realistic understanding of what is possible.

Adolescents may ask questions about whether or not their brother or sister will ever have a boy/girlfriend, will they get married, or drive a car, or live away from home.

As they get older they may start to wonder what role they will play in the ongoing care of the brother or sister. It is not uncommon for adolescent siblings to ask "Who's going to look after John when you're gone"? Clarifying their own, and their sibling's, future is an important issue for adolescent siblings.

If you try to ignore the questions or redirect the conversation because you're unsure of what to say, it can give the wrong impression to children. If you are unsure of the answer to a question, be honest, tell them you need time to think about it first and that you'll get back to them. Discuss the question with your partner, or a friend, get advice on the best way to answer. But always follow up with them to make sure that you have answered their question.

Having good open communication in the family contributes significantly to sibling well-being and adjustment. When you take the time to answer your child's questions, you are letting them know that disability is not a taboo subject and if they are confused or unsure they can ask and feel reassured.

**Mary Lloyd, Psychologist and presenter of the Association's Sibling Workshops for parents**

**From the September 2004 edition of NoticeBoard, magazine of the Association for Children with a Disability** www.acd.org.au **Tel 03 9500 1232 , Freecall 1800654013 (rural callers only)**

**Quotes**

"Questions about a sibling's difference aren't always meant to be unkind. Children learn about their environment by asking questions. Having a good understanding of their sibling's disability, and being prepared with a simple explanation can be very helpful when children ask the unexpected question."

"Finding the right words to explain a child's disability can be a challenge for parents. The trick is to keep it simple, but to give enough information that helps to explain the differences whilst trying to throw in some positives as well."

# UPDATE ON FRAGILE X RESEARCH

## Danuta Loesch

Dear all

Many families have been involved with our research studies in the past, and we are always grateful for families commitment and involvement in research into fragile X. I would like to update you on our work - and invite those who might be interested in participating in our studies to contact me for more information.
Our own research into fragile X is continuing on several fronts in collaboration with the team from the University of California at Davis, and our collaborative research is supported by the National Institutes of Health, USA (NIH).

**AUTISM** One aspect of our studies is to assess behavioural changes in fragile X, both in people carrying full mutation and premutation. We are especially interested in autism-like behaviours, and we are now re-testing earlier participants, and testing new participants using the latest (gold standard) tests for autism. The results of our most recent study have shown that more than half of full mutation boys, and a quarter of full mutation girls manifest autistic behaviours, and this is also true for some proportion of the carriers of premutation. We would like to understand more about the nature of these manifestations, and also assess how they relate to other behavioural and emotional problems that a person may have.

Moreover, my collaborator from the same Department, Dr Cheryl Dissanayake (a psychologist), is an expert in the early identification of autism, so now we also include infants, toddlers and young children with fragile X in our study. Identifying autism in young children is very important for families so that intervention may be implemented early in the child's development in order to maximise developmental outcomes.

**FRAGILE X-ASSOCIATED TREMOR/ ATAXIA (FXTAS)** is

another major aspect of our study. This newly discovered syndrome occurs in about half of older men carrying the fragile X premutation, and also in some proportion of premutation females as they get older, which of course is of concern. We conduct full neurological examination in these carriers, magnetic resonance imaging (MRI), and psychological and blood tests. Our preliminary results have confirmed a high prevalence of this syndrome amongst older males with premutation, but these data also showed that MRI changes may occur some time before the clinical manifestations,which may be important for early diagnosis, prevention and treatment should it become available. However, we still need more participants firstly, in order to confirm our preliminary results in larger samples, and secondly, to discover why some carriers develop this condition and some are symptom-free, and to identify the factors that may predispose or prevent this syndrome from occurring. We also need to establish at what age this disorder really begins. Therefore, we now need to include premutation carriers of all ages in our study, irrespective of whether they manifest any problems or not. We would also like to hear from all female carriers of premutation who may experience some relevant problems.

We have published a number of scientific papers from our fragile X study directed to professionals, and they are available through professional journals. For the readers of this Newsletter, however, viewing the website: www.FragileX.org is strongly recommended. This site is constantly up-dated based on our results, as well as on studies of many other researchers around the world.

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