

**KEEPING
IN
TOUCH**



**April
2006**

Cornelia de Lange Syndrome Association (Australasia) Inc.

Authority to Fundraise: NSW CFN 15201
Donations of \$2 and over are Tax Deductible
DGR 419321 ABN 97 070 990 653

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Websites: Australasia: www.cdlsaus.org
International: www.cdlsworld.org

Online support groups: OZ/NZ online discussion group: oznz-cdls@yahogroups.com
(register via the link on the www.cdlsaus.org homepage)
International Online Support Group: www.cdls-support.org

DISCLAIMER

This newsletter is not intended for diagnostic purposes or self treatment. The Cornelia de Lange Syndrome Association and its committee do not necessarily endorse or recommend any products, services, methods or literature mentioned within. Any questions about treatments should be discussed with your child's doctor.

From the Editor

In this issue, you'll find information on lots of coming events: the CDLS National Conference in Surfers Paradise, the International Conference in Canada, the Queensland Picnic Day and the NSW Adult Clinic Day. There's also a report from the Families BBQ in Canberra and an important article on gastro oesophageal reflux.

And a huge THANK YOU to Jenny from all of us for the medical folders that she compiled with help from Brett. It was an enormous job to write, photocopy, collate and post this fantastic resource to families.

Wanted: CdLS parents who speak Chinese (Mandarin) to talk to a new family from Taiwan. We are using an interpreter service to meet immediate needs, but this is a rather formal approach. We also need written information in Chinese if that is available. Please contact Jenny if you would like to help.

New Members:

Welcome to our new members from Queensland, NSW, ACT and Taiwan.

Donations:

Sincere thanks to the following people (and some not named here) who have financially supported the Association:

General: Japanese CdLS group

Special:

Palmer House, Wenona School, North Sydney. 2005 fundraising \$1,360.00 – used for printing and postage of the Medical Records Folders and Behaviour Questionnaires.

\$500 was donated so that the CdLS Association could contribute towards the purchase of a special set of scales for the adult disability clinic used by our families. Two other support groups who hold clinic days at this clinic also contributed.

In Kind: ComputerShare for printing and postage of KIT, printing of behaviour questionnaires.

Electronic KIT

The number of people opting for the electronic version of KIT is encouraging. If you would like to receive KIT this way just email Jenny on jennyrollo@bigpond.com with Electronic KIT as the subject.

Moving?? Please let us know your change of address when you move house. If you choose to receive KIT by email, remember to tell us if you change server.

CdLS National Conference 2006



“ *CdLS - Making Waves* “

Where: Hotel Watermark, Surfers Paradise

When: * Saturday 30th September 2006 – meetings, workshops
* Sunday 1st October – family fun day at theme parks or relaxing by the pool.

Speakers: Australia’s CdLS Medical Director, **Dr Meredith Wilson** (NSW) will update delegates about genetic research, **Dr Chris Oliver** from the UK will talk about Behaviours in CdLS, & Siblings Australia Director and author of *Siblings – Brothers and Sisters of Children with Special Needs* from South Australia, **Kate Strohm** will address siblings issues for parents and professionals in the general session, as well as hold workshops for siblings. Other workshops will be presented about issues for young people, and adults with disabilities – speakers to be confirmed.

Doctor Appointments: Will be available for Genetics and Behaviour.

Cost: We are applying for funding to help families attend, so hopefully there will be positive news soon. For now, the CdLS Association hopes to subsidise those of its members who are financial. We aim to cover the registration costs for the immediate family of such members. As is our usual policy, registration costs will be kept to an absolute minimum.

More details will be available soon. Registration papers will be sent in June.



Dr Meredith Wilson



Dr Chris Oliver



Kate Strohm

**International
CdLS Conference
July 2007**



**Niagara Falls,
Ontario**

“Niagara” is a native word meaning “meeting place”. How appropriate, then, that families and interested professionals from around the world will be meeting at Niagara Falls to learn all about CdLS. Set aside July 26th to 29th 2007 for the CdLS International Family Conference and concurrent CdLS Siblings Conference. The professionals’ two day conference will precede the family event. The Australasian CdLS Association expects to be well represented at this conference. More details as they are announced by the Canada Conference Committee. (Don’t forget to pack your camera!!)



Sunset and afternoon views of Niagara Falls – from the conference hotel!

2006 Qld Picnic in the Park

Please join us for our annual picnic at Orleigh Park, Hill End Terrace, West End on Sunday, 7 May 2006 from 11.30am. Pack your picnic basket, chairs, rugs and extended family and come along for a relaxing afternoon by the river under the shade of the Moreton Bay Figs.

For more info contact Rose or David on 3353 3470 or email

dave.humphrey@optusnet.com.au

Everyone Welcome!

CdLS Adult Clinic Day - NSW

The next CdLS clinic day will be held on 13th September 2006. At this time follow up appointments can be made. As at each clinic day there is time for two new referrals. New patients have an extended appointment in the morning where Dr Jane gives a thorough examination and takes a full medical history of the patient. Already one of these appointments has been taken by a new patient so there is only one spare this time around. If you would like your adult son or daughter to take this appointment, or if your child needs a follow up appointment please contact Jenny.



Anna, Dr Jane & David

Queensland Centre for Intellectual and Developmental Disability

The Queensland Centre for Intellectual and Developmental Disability (QCIDD) (known previously as the DDU), is located at the Mater Misericordiae Hospital in South Brisbane.

The QCIDD aims to improve the health and well-being of adults with intellectual and developmental disabilities, who live in Queensland. This is achieved through clinical practice, education, research and, more recently, advocacy. It also provides a tertiary comprehensive health assessment service for adults with an intellectual and/or developmental disability.

Clinic consultations occur at different times during the week, depending upon the clinician. Although anyone can refer to the QCIDD Clinic, **a referral from the person's GP is required**. A pre-appointment questionnaire will be sent to all persons referred to the Clinic.

Telephone: 3840 2412

Email: qcidd@sph.uq.edu.au

Website: <http://www.uq.edu.au/qcidd/index.html>

CDLS Families BBQ in Canberra

Four families with a CdLS member gathered on a sunny day in January for a BBQ in Canberra. Shane and Nolita drove up from Cooma with their little girl, Kiahna (CdLS 11 months) to meet with Christie and Chloe (CdLS 11 months). Chloe's sister Tayla, her grandmother Connie, and her Aunty Sam also came to be with us. Chloe's dad unfortunately was not able to come. Geoff and Meryl and their three daughters Amelia, Georgia (CdLS 6 years) and Ella also enjoyed the day. All the families provided wonderful goodies to share at the BBQ which was held at Phyl and Peter's home. Their son John (CdLS 26 years) and his older siblings Jane and Clare (with her children Phillip and Jasmine) helped to host the day.

Besides the laughter and the stories, the family focus of the day was enhanced by the fact that Sam and Meryl were both expecting babies within a few weeks. We're thrilled to report the birth of two beautiful children - a girl for Sam and a son for Meryl and Geoff. There was much excited discussion about the possibility of some of us attending the National Conference on the Gold Coast in September.

Whilst the adults sat in the shade and caught up on issues around CdLS, early intervention and current sporting events, Tayla, Amelia, Jasmine, Ella and Phillip made the most of the water pistols and garden hoses to cope with the very hot weather. Georgia skated around so expertly with her walking frame that we thought she was in training for the Commonwealth Games. John decided that his CD of Zorba the Greek was more inviting than all the activity and sat next to the music box. We are looking forward to meeting up again after winter and seeing how much Kiahna and Chloe have grown.



Georgia Crane



Peter Crawford with Chloe Blahos

Queen's Baton Relay

At the time of writing this newsletter the Commonwealth Games are underway in Melbourne. How beautiful was the opening ceremony??!! The highlight for me is always seeing the athletes "march" into the stadium. They are always glowing with excitement.

This time around I felt that excitement long before the start of the games. Running in the Queen's Baton Relay was an incredible buzz. Sitting in the runners' bus and looking back on the flashing lights of the police escorts' bikes and support vehicles and waving to the crowds in the streets made me feel like a celebrity.

The baton was quite heavy due to all the technology it contained. It had to be held in a certain way so that the computer camera inside could record the passing scenes. For a person of my advancing physical age (I'll always be young at heart!) having a young escort runner who was happy to help carry the weight was an advantage. In hindsight I should have walked instead of jogging. It took a month for my back to recover from the shock.

I enjoyed meeting up with other runners and assorted celebrities (real ones this time) at the evening celebration in Parramatta Park. Of particular note were Duncan Armstrong and Raelene Boyle who were happy to chat with everyone, sign autographs and pose for photos. And despite my protestations of being nobody of particular importance, a man asked for MY autograph! That was weird.

Copyright prevents me from printing actual running shots, so for your entertainment here are a couple from my camera. I'm thinking of changing careers. From science to photo journalism.... or perhaps a more appropriate job description is "stalker"!!



Raelene Boyle



Jen with Duncan Armstrong at the QBR evening celebration at Parramatta Park

Medical Records Folders

At the end of February a Medical Records Folder was posted to every person who has CdLS on our mailing list. Generally this was to the residential address of each person, but in some cases it may have been to a sibling, or other primary contact – not necessarily a primary carer. I would ask that these people please pass the folder on to the most appropriate person to complete for their loved one.

I realised when sorting through the mailing list that there are some people I am not sure of their relationship to a person with CdLS. For this I apologise, but there is a way to help me avoid incorrect mailings in future. Included in this issue of KIT is a brief Profile Form which you can fill in and post so that I can complete your details on our records. If you prefer, you can email me the details. Thanks for your cooperation.

Are you a primary carer and don't have a folder yet? (I need you to fill out a profile because I didn't realize you were a carer!) Are you a professional who would like to use a version of this folder for other clients? You have been sent a folder, but you're not a carer? Please contact me.

These Medical Records Folders were sponsored by the girls of Palmer House at Wenona School, North Sydney. These thoughtful girls have donated their fundraising from 2005 - \$1,360.00 - to the CdLS Association, ensuring every CdLS person in Australia has a folder. Thank you, Palmer House girls!

New Zealand families' folders have been printed and posted by Carol and Liz.

Parents of adults with CdLS have said they wished they had this folder at the beginning. Don't let the backlog of information stop you from using the folder from now. I have put David's together, and even without having written up his history yet, his information is now neatly arranged and easily found.

You will perhaps have thought of something you'd like to add. Please use as you see fit. Let me know if you have a brilliant idea of another proforma to add. Others will undoubtedly appreciate your ideas too. One glaringly obvious missing subject is behaviours. Any available proformas just weren't right for our kids. We have solved the problem though. Please read on about the Behaviour Research Survey. **Jenny**

CdLS Behaviour Research

SAC behaviour expert Dr Chris Oliver (UK) is conducting research into behaviours of people who have CdLS. Australian and New Zealand families now have the opportunity to contribute to the growing body of knowledge of this aspect of the syndrome.

As in the case of the medical folders, every person who has CdLS has been sent a questionnaire. Please fill in the answers as fully as possible and return to Jenny, or in NZ to Liz & Carol, ASAP using the supplied stamped envelope. The first "do by" date was 1st April. One more bulk mailing back to Dr Oliver will be done on 1st May. You can still return your completed questionnaire after that date. All information is important to the

research program. BUT By returning them early Dr Oliver has offered to write individual reports about each child's behaviour and how their behaviours compare to others around the world. This personal report can then be placed in your medical records folder for future reference. Having such a report will make it much easier to note behavioural changes in the future and easier for your child's psychologist/psychiatrist to help when/if these changes occur.

Dr Oliver will also be presenting a report at the Surfers Paradise CdLS conference about his findings from this research.

Please fill in your questionnaires NOW. Thank you to all those who have already sent their questionnaires back. **Jenny**

* The following article is reprinted from UK Reaching Out February 2006 – with thanks*

Canada team publishes its ophthalmology research results



Dr Alex Levin

The most thorough research into ophthalmologic issues among people with CdLS has been published in Canada in a paper authored by Dr Alex Levin – the chairman of the CdLS global federation's SAC and his colleagues at the hospital for sick children in Toronto.

The team was headed by Tamara Wygnanski-Jaffe, with support from John Shin and Enza Perruzza, and Mohamed Abdoell.

One hundred and twenty individuals with CdLS underwent ophthalmic examination to ascertain the relative frequencies of oculofacial and ophthalmic abnormalities.

To ensure that the diagnosis of CdLS was correct, Dr Laird Jackson confirmed the diagnosis in each case.

Dr Levin said: "We confirmed the frequent findings of synophrys (99%), long lashes (99%), hypertrichosis of the brows (96%), ptosis (44%), epiphora (22%), nasolacrimal duct obstruction (16%), blepharitis (25%), and myopia (58%). In addition, we found peripapillary pigment (83%), and microcornea (21%), which have infrequently been mentioned in the literature"

In conclusion, patients suffering from CdLS have many eye problems, some of which are readily treatable and demand early ophthalmic attention. The paper warns that ophthalmologists examining

patients with CdLS should look for myopic refractive errors and, once signs of blepharitis have been treated, consider probing and/or intubation of the nasolacrimal system. Most patients with ptosis will require surgical correction.

Article taken from the UK web site, with thanks. First published in November 2003

Secret signs of reflux

People with CdLS may not show the 'normal' symptoms of gastro oesophageal reflux disease (GORD) but, according to a new study, the number of patients with the disorder is much higher than previously expected.

For many years the Foundation has urged parents to ensure a test for 'reflux' is done as a standard. Many have reported back that doctors have said there is no point as their child is not showing signs of reflux.

GORD, or reflux, is defined as an abnormal return of acid gastric material into the oesophagus because of the bad function of the cardias (the valve that is supposed to stop anything coming from the stomach into the oesophagus). Everybody in the general population has reflux at some time, with 6 per cent of all adults and 11 per cent of babies under one year showing excessive signs or a pathological disease (GORD).

Symptoms

The typical symptoms of excessive GOR usually recognised by GPs include vomiting, regurgitation, refusal of food and frequent airways infections. However, the new research shows that many CdLS people show atypical symptoms such as bruxism (teeth grinding), nocturnal agitation, hyperactivity, and self injurious behaviour (SIB).

Scientific Advisory Council (SAC) member, Dr Angelo Selicorni, revealed the results of the research in advance of publication to CdLS families at the Connections meeting at Kilkenny, Ireland last month. Selicorni worked on the research with Dr S. Luzzani, Dr A.Valadè, and Dr F Macchini in Milan, Italy.

Speaking at the meeting, Dr Selicorni said that many of the previous studies of people with CdLS being affected by GORD were on small numbers of patients. Only that conducted by SAC member, Anne Marie Sommer, looked at as many as 17 patients. The Italian research project involved a study of some 43 patients who have a confirmed diagnosis of CdLS. The doctors set out to answer three questions:

Is GORD really a frequent complication of CdLS in a large number of the people affected?

Is there any correlation between the CdLS phenotype and GORD?

How important are behavioural symptoms?

The study involved evaluation of 43 CdLS patients – 18 defined as classical and 25 as mild. The age range was from one month to 30 years and there were 18 males and 25 females. The doctors carried out a 24-hour pH probe study on 41 of the patients; they did a GI endoscopy on 43 patients and a GI X ray evaluation on 23 patients.

The results showed that two out of every three people tested showed signs of having been affected by GORD.

The chances of suffering from reflux were equal whether the CdLS person was affected mildly or classically. However, the results showed that people defined as classical were nine times more likely to have severe reflux problems and mildly affected CdLS people were three times more likely to show slight reflux problems.

"The incidence of GORD is quite similar in the two different classes, yet the severity of GORD is strictly related to the severity of the CDLS phenotype," Dr Selicorni said.

The research showed that while only 53 per cent of the people affected showed typical symptoms, such as vomiting, some 83 per cent showed 'atypical' symptoms, such as hyperactivity. Half also showed problems at night, with poor sleeping patterns.

The findings were reinforced by Professor Chris Oliver, who said that in his behaviour studies the research team had noted a number of children showing signs of hyperactivity, back arching and self injurious behaviour that subsequently could be seen as indicating reflux. "It is a serious problem that needs to be considered," Professor Oliver told families at Kilkenny.

Dr Selicorni and the team will be arguing that there is a strong correlation between the behaviour issues and GORD. They believe that this needs to be noted by other paediatricians and by carers.

The Italian doctors went on to see what happened if treatment was carried out on the people affected and their research will prove that medical and/or surgical therapy eases GORD and improves related behavioural symptoms dramatically.

Search

“A careful search for GORD should be performed in every CdLS patient but especially when he or she demonstrates behavioural symptoms,” Dr Selicorni told parents.

With the study group they found that 61 per cent (17 of the 28 CdLS people who showed they were affected by GORD) could undergo medical (i.e. drug) treatment. The remaining 11 patients had too many problems and required surgical intervention.

Dr Selicorni said that these 11 patients underwent surgical correction using the Boix-Ochoa technique.

They were six male and five female patients with an age range from one to nine years. Of the CdLS phenotype: nine were classical (82 per cent) and two mild (18 per cent).

Apart from one patient, all operations were successful. The one patient who had problems was suffering from long-term respiratory complications. These have now been solved.

“The surgical approach is safe and effective in solving these situations,” Dr Selicorni told parents. The Italian doctors have continued to monitor the patients and have noted continual improvements.

Difference in techniques

Dr Selicorni said that people with GORD in Italy underwent surgery with the Boix-Ochoa technique. This is a fundoplication operation (effectively where a wrap is put around the faulty valve to force acid back into the stomach). In the UK and the USA the Boix-Ochoa technique has been replaced by the Nissen Fundoplication since 1993.

In a study into the two techniques at Booth Hall Children’s Hospital Manchester, surgeon, Subramaniam R. Dickson, reported that in a study of 109 fundoplication operations anti-reflux surgery is beneficial in children with significant gastro oesophageal reflux, irrespective of their neurological status, although complications are more common in patients who are neurologically impaired. Nissen fundoplication is more effective and has fewer complications than Boix-Ochoa, the doctors said.

The key messages from Dr Selicorni, and reinforced by the CdLS Foundation, are:

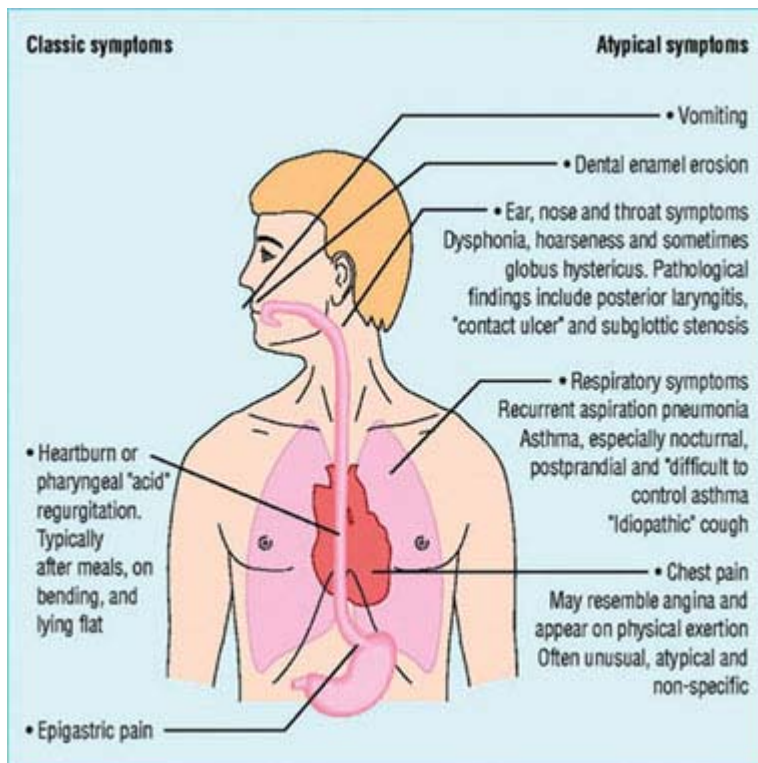
All CdLS children should be checked for oesophageal reflux even if they don’t show the normal symptoms.

Failure to treat reflux could lead to further problems, including ulcers and cancer of the oesophagus. This can be fatal, although it often isn’t.

Even mildly affected children can suffer from GORD but may not show symptoms.

All GORD can be treated either by drugs or by surgery. The surgery is proved to be safe.

- Literature about CdLS has often talked about GERD rather than GORD. This is because of the spelling of oesophagus. American spelling omits the first ‘o’.



UK doctor says don't panic

Leading UK consultant paediatric gastroenterologist, Peter Gillett, says parents shouldn't be panicked into unnecessary invasive tests to check out GORD.

"Tests are often unnecessary and a careful search for symptoms – typical or not – should be taken before any appropriate tests are done (pH, endoscopy or barium although the latter, as a test for reflux, is not great). Invasive tests are not desirable in any patient if they are not needed.

"My indication for fundoplication is failure of aggressive medical treatment or respiratory complications of reflux like pneumonia, if these don't settle with medical treatment."

Speaking at Peebles last year, Dr Gillett warned that fundoplication is not without complications. Problems include loosening of the 'wrap' and then patients need to go back onto medication. "There are number of CdLS parents who can attest to this," Peter says. There are also problems with 'dumping' – a bloating or retching against the closed valve at the junction.

Peter says the biggest problem facing parents is getting to see the appropriate person and recommends they get referred to a paediatric gastroenterologist.

Membership Form
CdLS Association (Australasia) Inc.

Date: _____ Enclosed is a Donation of _____

I wish to become an Active Member (Membership Fees \$25)

I wish to become an Inactive Member I wish to become an Associate Member (Professionals)

NAME: _____

Telephone: _____

Email: _____

Fax: _____

ADDRESS: _____

State: _____ Postcode: _____ Country: _____

PERSON'S NAME WITH CdLS: _____ Birthdate: _____

I am a parent Grandparent Relative Professional (incl. Field)

Other (specify) _____.

I wish to receive the KIT newsletter - yes / no

I give permission for the release of my name to other families for the purpose of mutual support:
yes / no

Signature: _____

Cheques made payable to CdLSA can be sent to The Secretary, P.O. Box 20 Putney, NSW, 2112, AUSTRALIA.

New Zealand Members only – Send \$NZ to 11 Winsomere Cresc. Westmere, Auckland 2 New Zealand

CdLS Association Profile

Name: _____

Postal Address: _____

Email Address: _____

I would like to receive KIT by post _____ email _____

I am a: Professional _____ field _____

Parent: _____ Grandparent: _____ Sibling: _____ Friend: _____

Other: _____

Name of person/s with CdLS: _____ DOB _____

_____ DOB _____ DOB _____

Siblings: Name/s and DOB

I give permission to release my name to other families for the purpose of mutual support: yes/no

Signature: _____ Date: _____

Return to: The Secretary, PO Box 20 Putney, NSW, 2112 Australia
