

**KEEPING
IN
TOUCH**



**MAY
2008**

Cornelia de Lange Syndrome Association (Australasia) Inc.

Authority to Fundraise: NSW CFN 15201
Donations of \$2 and over are Tax Deductible
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Websites: Australasia: www.cdlsaus.org
International: www.cdsworld.org

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

From the editor

This KIT includes reports and photos from the Adelaide clinic day, the Queensland picnic day is on 25 May (that's *soon*) and our major fundraiser is planned. There are also treatment protocols to assist families and professionals. A warm welcome is extended to our newest members from NSW.

Joanne

**It is time to renew your membership of the CdLS Association for the 2008/2009 financial year.
The cost is \$25 per year. A membership form is included in this KIT.**

Thank you to the following people for their generous support.

Donations – NSW: Jackson Family, J & D Watkins. Tas: Burston Family. SA: Routley, Scheer, Bednarz Families. WA: Sandilands Family. Qld: Humphrey Family. ACT: Crawford Family

Day of Disability – Rose Humphrey raised \$500 through an awareness day at work.

Printing and Postage of KIT - ComputerShare

WEB Sponsors – Humphrey & Rollo Families

ASSOCIATION NEWS

For further details about the following please contact Jenny Rollo.

Photos Wanted.

We are looking for photos of your CdLS loved ones to grace our new edition of the CdLS brochure. Please send good quality photos in jpg format by email to jennyrollo@bigpond.com.

While you are hunting through your photos you might like to update your space on our website gallery. You can also add a few words on what your child is doing now. In this case please send your good quality jpg photo and story directly to our webmaster, Rose Humphrey.

Collis Curve Toothbrushes are again available. Cost: one free to financial members, all others \$5 each, includes free postage within Australia. Adult and child sizes in various colours, all in medium hardness are available. These brushes help control gingivitis and make thorough brushing a faster, more pleasant experience. Very highly recommended!

NSW CdLS Clinic

Dr Jane Law has made time available specifically for adults with CdLS to have thorough health assessments. In March and September each year you can book a two hour consultation (for new patients) or a one hour follow-up appointment. Families who have used this vital health service have found great improvement in their child's health. Very highly recommended.

Appointments are possible at other times as necessary.

Consider joining the management committee. You can help in the running of our Association by deciding to join the management committee. Meetings are held by phone 3 or 4 times a year, usually on a Saturday afternoon. You need to be a financial member to apply.

Canada 2007 - DVD Rom of International CdLS conference presentations is now available.

QUEENSLAND ANNUAL PICNIC DAY

Where: Orleigh Park, Hill End Terrace, West End

When: Sunday, **25 May 2008**, from 12 noon onwards

Directions: Come down Montague Road, West End and turn right into Hill End Terrace. We will meet at our usual spot near the play equipment, if space is available, otherwise just look out for the group with the CdLS Banner.

I hope to see you all there.

Rose

Major fundraising Event

For the first time in eight years, the Australian CdLS Association is holding a fundraising event. This time around we will enjoy an evening of theatre. Bernadette Feeney (Dalingwater) is a RADA trained actor playing the title role of Shirley Valentine by Willy Russell. This one-woman play will have you enthralled! Bernadette is the sister of Michael Feeney aged 52 years who has CdLS, and also the President of CdLS Canada.

About the play: See the flyer included in this mailing.

The challenge is on for families who live interstate, and in country NSW to become involved. If you are unable to travel to Sydney to attend, perhaps you know people who can! Ask them to book a group of their friends. Pass on the flyer. Or maybe you and your contacts would prefer to donate. All help is welcome.

All money raised goes directly into projects which help our families. We have no salaries, minimum administration costs, etc because we are a non profit organisation run entirely by volunteers.

The Shirley Valentine flyer is available in PDF for emailing to your friends and family.

Shirley Valentine

Friday 12th September 2008 7.30pm

Riverside Girls High School Auditorium

Huntley's Point Road, Gladesville. (Near Gladesville Bridge, **SYDNEY**)

Tickets \$65 each - Includes wine and nibbles.

Group Bookings: Buy nine tickets, the tenth ticket is free

Phone (02) 9809 0287 after 3.30pm weekdays. We accept Visa, Mastercard and cheques.

An auction and raffles will also be held on the night only for those in attendance.

Drugs used to treat aggression not working –

Sydney Morning Herald - January 5th 2008

DRUGS that are commonly used to treat aggressive or violent outbursts in intellectually disabled people are less effective than a placebo and should not be used as a standard form of treatment, research shows.

The finding, by Australian and British experts, strongly challenges routine medical practice throughout the world of using antipsychotic drugs to treat aggression in intellectually disabled patients. Up to 45 per cent of people with an intellectual disability in hospital and about 20 per cent of those in the community are prescribed antipsychotic drugs, although there is no clear connection between aggressive behaviour and psychotic illness.

The study, published in *The Lancet*, examined 86 adults with a mild intellectual disability in group housing in England, Wales and Australia over more than a month of treatment. It found a 79 per cent reduction in aggressive behaviour among patients taking placebo pills, compared with a reduction of 65 per cent or less in those taking antipsychotic drugs.

Researchers compared the placebo with two antipsychotic drugs - haloperidol and risperidone - although the findings would almost certainly apply to all similar medications, they said.

The lead author, Peter Tyrer, a professor of psychiatry at Imperial College London, said that although all treatments led to a reduction in aggression after four weeks, the greatest decrease was by those taking the placebo. "Our trial has shown that aggressive challenging behaviour in people with intellectual disability decreases whether or not active medication is given," he said.

There had been no differences between drugs and dummy pills when measuring aggressive behaviour, quality of life, effect on carers and adverse drug effects, Professor Tyrer said.

The study's authors, including researchers from the University of Queensland, said the results "should not be interpreted as an indication that antipsychotic drugs have no place in some aspects of behaviour disturbance".

Dr David Harley, who worked on the study while at the Queensland Centre for Intellectual and Developmental Disability, said he was not surprised that the drugs had little more effect than the placebo, given they had not been used for the purpose in which they were created. "They are being used to treat [aggression] which is not a recognised medical diagnosis," he said. "We might expect drugs like this to work if the aggression was caused by schizophrenia or psychotic illness."

Dr Harley said when intellectually disabled people became aggressive, doctors were left to feel "like the only avenue they have is to prescribe". He has been advocating against the use of medication in this group for years and preferred to treat most patients with behavioural therapy.

Philip Mitchell, head of the school of psychiatry at the University of NSW, said the study was a "wake-up call" to psychiatrists that the drugs were "of limited benefit" for patients with intellectual disability. "It should hopefully make clinicians and doctors more circumspect about their prescribing practices," he said.

A note from Jenny: David had been taking Risperdal for aggressive behaviour over a period of about 3 years. Since I have weaned him off it his behaviours have improved enormously, and he is far more aware of, and involved in, his surroundings. However, some people with CdLS do benefit from using this drug. This news article was included in KIT for information purposes. Always consult your doctor for individual advice.

ADELAIDE CLINIC DAY

Thirty people attended the most recent CdLS meeting – a clinic day held in Adelaide in February 2008.

Two very dedicated doctors donated their time and energy to meet the complex health needs of our CdLS children.

Prof. Chris Oliver – international SAC expert on behaviours from the UK presented his “behaviour plus” talk. He saw patients during extended consultations. Chris also did a home visit while he was passing through Sydney to see a child with CdLS who was unable to travel to Adelaide. This was very much appreciated by the parents.

Dr. Jane Law – presented case histories and discussed many medical problems to look out for in CdLS during her talk to the delegates. This was particularly appreciated by parents of the younger children, as they now feel better prepared to advocate for their child. She worked very hard doing extended consultations and preparing full reports for patients. Parents are now following up with local doctors. Jane had also researched resources in their home states for parents.

Thanks to **Jemma Rollo B. Psych (Hons)** – who acted as Chris’s note taker during the behaviour consultations. Chris found this very useful as he was able to concentrate on talking to the parents, making best use of the time allocated to each patient.

At this meeting we decided to limit the formal proceedings to two major areas of concern to families – behaviour and general health issues. This allowed for longer consultations with the doctors and more time for parents to talk with each other in a relaxed atmosphere. We are encouraged to again use this more relaxed format for future meetings.

Special thanks to **Lil Routley** (mum of Ella, 2yrs) from Adelaide who was instrumental in the organising and smooth running of this event.



Prof Chris Oliver (UK) and Dr Jane Law (NSW)



Colin, Sharyn & Kate Burston 17 (Tas) with Lil Routley (SA)



Keith, Jane, Ben (28) & Tim (SA)



Jodie (36) & Judy (SA)



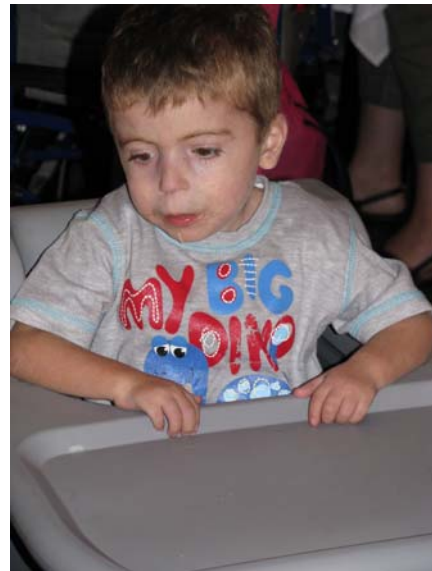
Yvonne, Carlo & Gia (4) (SA)



Corey (4) (VIC)



Ella Routley 2 (SA) and Daniel O'Rourke 3 (NSW) enjoyed their time in childcare



Daniel O'Rourke 3 (NSW)

AND THE WINNER IS *BRIGHTON!!!*

The **International CdLS Conference** will be hosted by the United Kingdom and Ireland in **2009**.

Start saving and planning your side trips! The conference will be held in **Brighton, England**.

Professionals and federation leaders – **22nd and 23rd July 2009**

Families – **24th to 26th July 2009**

If our gossipy women's magazines are right we may be there in time for another royal wedding!

Krystal's Braces by Susan Gerrard, mum of Krystal (CdLS) 14yrs.

In December 2003 we were told that Krystal needed braces. We rang up the Dentist every 6 months but were told every time "another 3 years" because of the waiting list.

After August 2007 I spoke to Krystal's doctor about her teeth. He offered to write a letter to the dentist and also get a letter from her school speech therapist. In only two weeks we had scheduled our first appointment, and 10 days after that the braces were on.

Since then we have 6 weekly visits. Krystal needed 2 teeth pulled out. Now she is wearing little rubber bands 24 hours a day, takes them off for meals, then she cleans her teeth and puts the bands back on.

We are very proud of Krystal. She has been very brave, especially when the teeth were pulled out, and then when the braces were put on.

The dentist said she will need to wear the braces for 12 to 18 months.



Krystal's now stunning smile.



Krystal's teeth before treatment



Treatment Protocols

The following treatment protocols are from the website of CdLS UK and Ireland. More protocols are available at <http://www.cdls.org.uk/treatment/index.htm>

Puberty

Puberty appears to be a difficult period for many individuals with CdLS. The onset and course of puberty appears similar to unaffected individuals, not only physically, but also emotionally and behaviourally. Some characteristics have included mood swings, irritability, unexplained pain episodes, contrariness, worsening behavioural problems, and aggression. In females, premenstrual syndrome occurs and may be treated symptomatically (e.g. Tylenol or non-steroidal anti-inflammatory agents). Menstruation may be a problem for caretakers, especially in females with communication difficulties. In addition, protection from potential pregnancy may be indicated, since for both males and females fertility appears to be normal or slightly decreased. Several therapeutic modalities have been utilized including synthetic hormonal treatment (e.g. oral contraceptives or depo-provera injection) and surgery (e.g. tubal ligation, hysterectomy). Together, the individual with CdLS, the family and the practitioner should determine the most appropriate course of action.

Undescended testicles are certainly at increased incidence in males with CdLS. Nearly 10% of affected males have one testis or both testes undescended, compared with less than 2% of the unaffected male population. In general, surgery is definitely recommended following failure of medical treatment (hormone injections) to bring down the testicles. The primary reason is because testicles that remain in the abdomen are at higher risk for developing malignancies than those that are in the scrotum, even though this is a fairly low percentage.

The secondary reason is for the fertility issue, which may or may not be a factor in an individual with CdLS, depending on the level of function. The earlier the procedure can be done (e.g. by age 2), the less are the psychological effects of the surgery, but it should be performed prior to puberty, since that is when the risk for cancer rises.

Speech and Language

One of the most challenging issues for caretakers or people with CdLS is determining the best communication tool available for their use: speech, signing. Communication boards, computer etc. While it is not clear why some children talk and others do not, particularly among the children without obvious differences in physical appearance, it is very clear that all individuals do communicate.

For some people with CdLS, speech develops normally. For most, however, the ability to communicate is greatly influenced by other developmental factors as well as access to early intervention programs and speech therapy. Children who weighed at least 5 pounds at birth, who had no or mild hearing loss, who had no severe upper-limb malformations, who sat by the age of 18 months or walked by the age of 30 months and who were judged to have good social relatedness were much more likely to acquire expressive language skills than those who did not meet these criteria. Social relatedness included factors such as eye contact, the appearance of comfortableness, alertness, and the child's overall ability to relate to people.

Characteristics of Speech, Language, and Hearing

The absence of speech or the development of only minimal speech has been well documented, even in the more mildly affected. There is often a characteristic vocal quality in the crying of babies, which has been described as feeble or low-pitched. The low-pitched cry frequently present at birth and early infancy often disappears by 12 months.

Most people with CdLS exhibit errors in articulation. Consonants are typically distorted or missing. In addition, there have been some reported observations of severe oral-motor and verbal apraxia, which is the loss of the voluntary aspect of speech and motor movement.

There have also been unconfirmed reports that individuals have a tendency to unexpectedly utter a meaningful word or phrase only once, using completely clear articulation and often performing at a level higher than previously observed. But then this performance is rarely if ever repeated.

In almost all individuals the ability to produce language was remarkably inferior to the ability to comprehend language. There was also considerable discrepancy between vocabulary measures and syntactic skills in that people who had highly developed vocabulary usually did not exhibit the expected syntactic skills. Similarly, children who were using an average utterance length of 4-5 words per utterance typically were not using question transformations.

The majority of individuals are very quiet. They often make eye contact and there is often a sense they understand what is being said, but they are not usually vocal. Even among individuals who have good language skills, there are few who can be described as talkative.

The decision to begin speech therapy should not be delayed. Some parents have reported they have been told speech therapy could not begin until their children were talking! Additional parents reported they were told their children could not receive speech therapy or learn to talk until the gastric tube was removed and their children were eating normally. This of course is not true.

Some children who are talking have never received nourishment except through their gastric tube. However, when appropriate it would be beneficial to work with a speech-language pathologist on feeding therapy so the oral mechanism functions as normally as possible.

Augmentative and Alternative Communication

For some people with CdLS, non-vocal or non-verbal strategies will need to be considered. For these individuals it may not be possible to achieve normal or even adequate speech for communicative purposes. Instead the ultimate goal should be to communicate at a level adequate to meet communication needs. Examples of available strategies include a communication board; American Sign Language; American Indian Hand Talk or Amer-Ind gestural code; Blissymbolics; Total Communication; Pantomime; a manual alphabet; eye-blinking encoding; or electronic communication aids.

For children with severe upper-limb malformations there are gestural-assisted and neuro-assisted strategies available. Unfortunately almost all augmentative communication strategies are difficult to learn for children who have difficulty understanding nonverbal or gestural communication. Regardless of the disability, it is important that all individuals be taught some means of indicating "yes" and "no."

Before considering the use of augmentative or alternative communication, a number of factors need to be considered. These include the level of cognition, motor abilities, receptive language abilities, and the motivation to communicate. It will often be most beneficial when speech-language pathologists function as communication therapists rather than as speech therapists and consult as appropriate with professionals such as physical therapists, occupational therapist, physicians, psychologists, social workers, vocational counselors, nurses, and teachers.

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.