

KEEPING
IN
TOUCH



JULY
2010

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@yahoo.com
(register via the link on the www.cdlsaus.org homepage)
International Online Support Group: www.cdls-support.org

From the Editor

Many thanks to Jenny Rollo for putting KIT together last issue while my daughter Brodie was in hospital for surgery for a twisted bowel. I've written a bit about our experience in this issue of KIT in case it is useful.

The AGM is coming up in September, held physically in Brisbane and by phone from wherever you are! Please consider nominating for a position as a member of the committee, which meets by phone. It is not time consuming and your ideas and opinions would be very welcome. Membership and Nomination forms are at the end of this KIT.

Joanne

Membership News

Membership Fees Paid: NSW – Freeman, Rollo, Yapp families, S. Elliott. QLD- Gorman, McNamara families. WA – Arnett family. VIC – Eisele family. Malaysia – Nor family.

Donations: M. Nuttall, E Chipman, F. Yapp., M. Tudor, M. Nor.

Donations by Regular Deduction: S. Kinchela

Welcome to new families from NSW and Victoria.

Notice of Annual General Meeting

25th September 2010

11am

in Brisbane at the Marque Hotel

or by telephone

If you wish to attend by telephone conferencing please contact Jenny Rollo by 15 September jennyrollo@bigpond.com

Prof Chris Oliver goes to any length to help people with CdLS.



This UK summer Chris Oliver cycled the length of Britain raising over £3,800.00 for the UK CdLS Foundation. From Lands End to John O'Groats Chris not only cycled, but kept up a very amusing blog and Facebook updates for his sponsors and fans. "Chris could get a second job as a comedian", said one Australian fan."I was in hysterics reading his blogs!"

Perhaps we'll let him do the comedy club rounds when he retires. For now we need him to continue his essential work as our CdLS behaviour expert.

Congratulations, Chris -an amazing feat. We look forward to welcoming you (and those well-toned legs!) to Brisbane for the CdLS Clinic day.

Queensland News



Back: Laura and Krystal Centre: Katie Front: Isabella

At the recent QLD **CDLS** mid-year picnic the families that attended had a great day (although it was very cold) and the **CDLS** children were very well behaved. We all noticed a big difference in our beautiful children.

During the day the adults discussed future picnics and it was decided that our midyear picnics would be held each May (last Sunday) at Orleigh Park and our Xmas Picnic would be held on the 2nd last Sunday of November at the home of John and Margaret Wood. Information about our 2010 Xmas get together is included in this edition of KIT (previous page).

Attending families at these get togethers always enjoy the day and we would encourage more families to take the time to come and join us.

By Margie Wood (sibling)



CDLS CHRISTMAS PICNIC – 2010

Come and join us for our 2010 Xmas Party

WHERE

John and Margaret Wood's Place
289 Old Cleveland Rd East
Capalaba, 4157

WHEN

Sunday 21 November 2010
10.30 – 3.00

A sausage sizzle lunch and cordial will be provided

BYO nibbles, chairs and drinks

All children will receive a present

RSVP - Margaret on 3823 2245 by Monday, 15 November

A good time will be had by all

See you

Margaret Wood

Twisted Bowel

In April this year, our 19 year old daughter Brodie became very upset with strong abdominal pain and some vomiting. She has wind pain every day, but this was different. She is non-verbal but her distress was clear and she was 'straining' as if constipated, but wasn't. On our doctor's advice, we went into Emergency at a large public hospital.

An x-ray showed no blockage and she improved as we sat there, so we went home. A fortnight with no problems and suddenly the pain was back. Again into Emergency but this time the x-ray showed a blockage, confirmed by CAT scan (under general anaesthetic) and she went straight into surgery and had that part of her bowel removed. A part of the bowel had twisted on itself and became blocked (this is called a volvulus). Apparently, the bowel can twist and then partly untwist, which explains the pain – normal - pain sequence she had.

A twisted bowel is different to a bowel malrotation, which is a birth defect sometimes found in CdLS. Brodie does not have a malrotated bowel.

We were in hospital for a fortnight. She's back to a normal routine now but still needs a bit more time to be fully recovered.

Perhaps this might help another family presented with similar symptoms – something to at least mention to the doctor.

Joanne Argent

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.

CORNELIA DE LANGE SYNDROME

Clinic Day

Brisbane, Queensland

Saturday, 25th September 2010

The Conference Venue

The Marque Hotel, 103 George Street, Brisbane ... is located in the heart of the city, adjacent to the Treasury Casino and just a minute's walk to the Brisbane River. The hotel's central location offers easy access to the best that Brisbane has to offer - cruising the Brisbane River, shopping along Queen Street, soaking up some sun at South Bank Parklands, and cheering till you're hoarse at iconic sporting venues such as The Gabba. As well as the riverside walks, the hotel is easily accessible to Queensland University & Botanical Gardens, the Queensland Art Gallery, Performing Arts Centre, the Brisbane Convention and Exhibition Centre.

- ✓ Deluxe Rooms – up to 2 adults and 1 child (rollaway bed) - \$159 per night
- ✓ Buffet Breakfast – \$10
- ✓ Deluxe Riverview Rooms, Executive and Spa Suites also available.

When making accommodation reservations please let them know that you are with the Cornelia de Lange Syndrome Conference. There are limited disabled rooms, so book early to avoid disappointment. Reservations: Jina or Sean Tel: (07) 3221 6044

FROM	DISTANCE	TIME	TRANSPORT
Airport	15 km	25 min	Car
Roma Street Station	0.9 km	10 min	Foot
Downtown	0.5 km	5 min	Foot
Queen Street Mall	.05 km	5 min	Foot
Treasury Casino	.05 km	5 min	Foot
Southbank Parklands	1.5 km	15 min	Foot

How to get to the Marque Hotel ...

From Brisbane Airport take the Airtrain from Brisbane Airport to Roma Street Station – exit the station and head south-west on Herschel Street, turn left at George Street, hotel is approx 6 blocks on the left. (Airtrain timetable and fares www.airtrain.com.au)

It may be cheaper and more convenient for large groups to travel by taxi from the airport to the hotel, except in peak hours when traffic around the airport can come to a standstill for lengthy periods of time.

Local residents should be aware that there is limited parking in Brisbane CBD. The Marque Hotel offers secure undercover parking for residents at \$20 per day.

Childcare will be in the capable hands of the Everton Park Girl Guides.

About the Presenters:

Chris Oliver, BSc, MPhil, PhD, AFBPsS, CPsychol – Professor of Clinical Psychology at the University of Birmingham and trained as a clinical psychologist at Edinburgh University before completing a PhD on self-injurious behaviour in people with intellectual disability at the Institute of Psychiatry, London. He is currently researching behaviour disorders in people with severe intellectual disability and gene-behaviour associations and is on the Scientific Advisory Committee of the Cornelia de Lange Syndrome Foundation with a particular interest in behaviour disorders.

Dr Barry Lipschitz – Paediatric Gastroenterologist, Melbourne has treated a number of patients with CdLS for gastrointestinal disorders. He will be presenting his research findings of gastrointestinal problems in CdLS.

Program

Friday 24th

Evening – Time TBA – Management Committee Meeting

Saturday 25th

8.30am Registration for Families and Professionals

9.00am Childcare opens. Please collect your children promptly for morning tea and lunch.

9.00am Behaviour Challenges in CdLS - Prof Chris Oliver (UK).

Gastrointestinal Issues in CdLS – Dr Barry Lipschitz (Melb.)

10.30am Morning Tea

11.00am Doctor consultations begin

11.00am CdLS Association (Australasia) Inc. AGM

12.30pm Lunch

1.30pm Consultations continue

3.00pm afternoon tea

6.30pm Dinner

Sunday 26th

8.30am State Coordinator Training

Noon. Meetings end.

PROFESSIONALS REGISTRATION FORM

Cornelia de Lange Syndrome Clinic Day

Saturday 25th September 2010

Surname: _____

First name: _____

Title (Dr, Ms, Mr, etc): _____

Professional field: _____

Address: _____

Telephone: _____

Email address: _____

NOTE: Professionals who are financial members of the Association receive Keeping in Touch newsletter and **free registration** to this seminar for one person.

Date	Time	Event	Number attending	Cost Per person	Total Cost
Saturday 25 th September	9am to 11am	Seminar (Includes morning tea),		\$25 or one free to members	\$
I wish to become a Financial member of the CdLS Association (optional)				\$25	\$
					\$

Do you need a parking space? Yes / no

Select Payment Method (please tick)

Cheque enclosed (payable to CdLS Australasia Inc)

or

Please charge my Visa / Mastercard account

Card number _____ Expiry date _____

Name on card: _____ Signature _____

Please read and sign Release Form

FAMILIES REGISTRATION FORM

Cornelia de Lange Syndrome Clinic Day

Saturday 25th September 2010

General information

Family name: _____

Address: _____

Telephone: _____

(include area code)

Email address: _____

Family members

1. Name of family member with CdLS _____ Age _____

Is s/he attending? _____

2. Please list other members of your family / group attending the Conference. Names should be as you wish them to appear on name tags.

ADULTS attending the Conference

Family name	First name	Relationship to person with CdLS
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CHILDREN attending the Conference

Family name	First name	Relationship to Person with CdLS	Age
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Special Requests

1. Do you require a highchair? _____ If so, how many? _____

2. Does anyone in your family have special dietary requirements? _____

Do you need a parking space? Yes / No

Accommodation

Are you staying at The Marque Hotel? Yes / No Delegates please make your own bookings with the hotel

Optional Dinner

Delegates are encouraged to attend the conference dinner. Previous experience has shown this to be the highlight of the social side of our gatherings.

Doctor Consultations

Two specialists and a general practitioner (GP) will be available for individual consultations with families on Saturday afternoon. Sessions will be about 20 - 30 minutes long. You can request a consultation even if your CdLS person is not at the clinic day.

INDICATE WITH A number 1, 2 and/or 3 in order of importance YOUR REQUESTS FOR CONSULTATIONS:

- ____ Behaviour
- ____ Gastroenterology (Bring your Medicare Card with you)
- ____ General Health (Bring your Medicare Card with you)

Please read and sign Release Form

Liability, Information and Photo Release

All adults attending the 2010 CdLS Clinic Day must sign their consent to the following:

I release CdLS Association (Australasia) Inc., committee members, officers, and any other person acting on behalf of or acting or purporting to act as agent of CdLS Association (Australasia) Inc. including volunteers, from any liability whatsoever arising from any conduct including any alleged negligence by act or omission of CdLS Association (Australasia) Inc. and of any other person acting on behalf of or acting or purporting to act as agent of CdLS Association (Australasia) Inc. in connection with my attendance, and the attendance of any member of my family, at the 2010 Clinic Day.

I give permission to CdLS Association (Australasia) Inc. to release my name to professionals participating in the 2010 Clinic Day.

I give permission to CdLS Association (Australasia) Inc. to use my name and image (photographs and/or video) as well as that of family members at the 2010 Clinic Day in any CdLS Federation publication.

Requires the dated signatures of each attending adult below:

Name _____ Signed _____ Date _____

Name _____ Signed _____ Date _____

PAYMENT FORM FOR FAMILIES

Registration Fees for Family Conference

There is a substantial discount for financial members of the CdLS Association. Financial membership costs \$25 per family per year. If you and your family are not currently financial members, you are only eligible for the discounted fees if you complete and return the enclosed Membership Form and payment with your Conference Registration Form. The cost for non-members is also being partially subsidized.

Registration Fees for Financial Members

(Limited to 2 parents per membership):

Registration Fees for non-members:

People with CdLS:

Adults Free **Siblings Free**
Adults \$50 **Children \$20**
Free

<u>Attendees</u>	<u>Number</u>		<u>Cost</u>	=	<u>Total</u>
Annual Membership of CdLS Association			\$25	=	\$ _____
Adults (max. 2 parents) _____	x	member		=	\$ 0
Adults _____	x	\$50		=	\$ _____
Children _____	x	member		=	\$ 0
Children _____	x	\$20		=	\$ _____
Person with CdLS _____	x	free		=	\$ 0
Payment Total					\$ _____

Registration includes: Lunch, tea breaks, childcare, stationery

Optional Dinner

Attendee Numbers

Adults _____ Children and CdLS _____

Numbers are for booking purposes only. **Each guest to pay for their own meals.** We estimate prices to be about \$50/head adults & \$25/head children.

Select payment method:

_____ Cheque enclosed (made payable to CdLS Association (Australasia) Inc.)

_____ Please charge my _____ Visa _____ Mastercard _____ account.

Card number _____ Expiry date _____

Name on Card: _____ Signature _____

Post registration forms to: CdLS Conference Registration

PO Box 20
 PUTNEY, NSW, 2112

Membership Form
CdLS Association (Australasia) Inc.

Date: _____ Enclosed is a Donation of _____

_____ I wish to become a Financial Member/renew membership (Membership Fees \$25)

_____ I wish to become an Associate Member (Professionals) (Membership Fees \$25)

NAME: _____

Telephone: _____

Email: _____

Fax: _____

ADDRESS: _____

State: _____ Postcode: _____ Country: _____

NAME and birthdate OF PERSON WITH CdLS: _____

NAME(S) & Birthdate(s) of siblings: _____

I am a parent _____ : Grandparent _____ : Relative _____ :

Professional (incl. Field) _____ :

Other (specify) _____ .

I wish to receive the KIT newsletter - by mail by email not at all (please circle choice)

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

Signature: _____

Payment Method:

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

Credit Card:

Mastercard _____ Visa _____ Bankcard _____ (Please tick)

Card Number _____

Cardholder's Name (Full name as it appears on card): _____

Card Expiry Date: _____ Total: _____

Signature of Cardholder: _____

Thank you for your financial support.

Note: If attending the Brisbane Clinic Day, Membership fees are included in the Clinic Day registration payment

The following article is from the USA web site Treatment Protocols – with thanks:

GROWTH AND DEVELOPMENT

Growth

Individuals with CdLS have proportionate small stature. Specific growth charts are available for height, weight and head circumference parameters of boys or girls with CdLS at different ages. Using these, the practitioner can follow growth as compared to other affected individuals rather than the unaffected population. If an individual deviates from the CdLS curve, then specific investigation for occult pathology may be warranted. The term "failure to thrive" is not appropriate, and may generate the use of unnecessary calorie supplementation, which results in no increased rate of growth and considerable parental anxiety. Pubertal growth spurts occur at comparable ages to unaffected individuals. Some individuals with CdLS have been found to have growth hormone deficiency, requiring exogenous growth hormone administration, however, without a documented deficiency, the use of growth hormone most likely does not increase final adult height.

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.

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.

Undescended testicles are at increased incidence in males with CdLS. Nearly 70% 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.

Growth Hormones

There is not much factual information available at this point regarding the issue of growth hormone testing and therapy. In the meantime, growth in CdLS can be monitored by using the CdLS specific growth charts. If growth is below the lower curve, then other factors may need to be taken into account (e.g. growth hormone deficiency). Several cases of growth hormone deficiency have been documented, probably higher than the incidence in the unaffected population, but this has

not been studied carefully. Certainly with documented growth hormone deficiency, replacement hormone may be appropriate.

Development

Developmental skills are often delayed in individuals with CdLS. Because of this, early intervention is indicated as soon as a diagnosis is made, since it appears to play a major role in the level of developmental achievement. An average IQ score in individuals able to be tested has been found to be 53, which is within the mild to moderate range of mental retardation. Acquisition of new skills continues throughout life without regression. Developmental areas of strength include visual-spatial memory and perceptual organization, and thus the use of computers, using visual memory, may be of benefit over standard methods of verbal instruction, as may tactile stimulation. Language is an area of weakness, and may be compounded by hearing abnormalities. Fine motor activities should be stressed in education, especially those related to activities of daily living. A developmental chart specific for CdLS is available, and can be used by practitioners and therapists to monitor progress, and potentially even to confirm diagnosis.

Thanks to the NSW Association for Children with a Disability for this article:

National Disability Insurance Scheme

- **It's time to change the way services for people with disabilities are funded and structured in Australia.**
- It's time to make services for people with disabilities and their families effective, equitable and efficient.
- Almost every Australian has cared, or is caring, for a family member with a disability, or knows of a family doing so. Many of these families fall through a huge gap in Australia's social services network.
- Those who acquire a disability through a workplace or a motor vehicle accident generally receive financial support. However, for those who acquire permanent disabilities in other accidents, are born with a permanent disability, acquire a permanent disability through a medical condition or have a permanent mental illness, there is no automatic support to meet their needs.
- This is a national disgrace and an issue for every Australian.
- It's time to introduce a new national safety net, ensuring peace of mind for all Australians should they or someone they love become severely disabled.
- In place of Australia's current crisis-driven, outmoded welfare-based approach to disability services, it's time for a modern, forward-thinking National Disability Insurance Scheme.
- Governments are already spending billions of dollars on disability services. A national insurance approach with its inbuilt focus on lifetime needs and claims management would be much more effective and efficient, as well as fair.
- With the number of Australians with a severe physical, intellectual and/or behavioral disability increasing, and the community's capacity to provide informal care declining, it's time to start planning properly for the future, as Australia did in the 1980's when compulsory superannuation was introduced.
- Its time to cement a National Disability Insurance Scheme as a central plank of Australia's social and economic policy framework.

The Plan

- **Australia's approach to disability services is crisis-driven and welfare-based. A National Disability Insurance Scheme (NDIS) would change this. The Scheme would provide funding for essential care, support, therapy, aids, equipment, home modifications and access to the community, education and training.**
- As an insurance-based scheme, providing cover to Australians as and when they needed it, an NDIS would be funded by all taxpayers through general revenue or an extension of the Medicare insurance levy. An insurance-based approach to disability services stems from the view that disability is a "risk" that can strike anyone in our society, with potentially catastrophic consequences if proper support services are not in place, but will not affect everyone.
- Because an NDIS would be a major social and economic reform, on a par with the introduction of Medicare and compulsory superannuation, the first necessary step towards implementation is a detailed feasibility study that would allow for careful, thorough planning and consultations with the States and all stakeholders.
- It is envisaged that principal beneficiaries would be those Australians with profound and severe disabilities (approx 700,000) who need assistance with daily living tasks (self care, communication and mobility) while people with more moderate disabilities could also be eligible for some assistance based on their lesser needs.
- The defining criteria should be needs, as determined by clear eligibility criteria, which would include measures of physical, intellectual and behavioral disabilities.
- It's estimated that only some 3% of the population would be the primary beneficiaries of an NDIS. But all Australians would benefit from the peace of mind of knowing that a Medicare-type safety net was in place, should they or someone they love be affected by disability, either from birth or as the result
- of accident or illness.
- Without a comprehensive funding plan to meet future need, Australia faces social catastrophe as an entire generation of ageing carers die, other carers and people with disabilities become increasingly isolated from the labour force and the community, and many more families collapse under the strain.

Four Ways to Take Action

Join the National Disability Insurance Scheme's rapidly growing national coalition of supporters in urging Australia's governments, federal and state, to **Take Action Now**.

Email your MP Tell your MP how an NDIS could change your life.

Visit your MP Visit your MP to tell them about NDIS in person.

Participate An inquiry into a long-term care and support scheme for people with a disability (a National Disability Insurance Scheme) has been announced and you can be part of it. Please visit www.pc.gov.au/projects/inquiry/disability-support and register your interest.

Spread the Word Become a Facebook fan, Twitter the message, circulate the petition, request a campaign email to send to friends or encourage your local newspaper to do a story.

Cornelia de Lange Syndrome Association (Australasia) Inc

**Nomination Form For Position of Office Bearer or Ordinary Committee Member
in The Above Mentioned Association**

I, _____

Of (address) _____

_____ STATE: _____ P/CODE _____

and being a current financial member in good standing of the above mentioned

Association, do nominate _____ for the position of:

signed: _____ Dated: _____

I, _____

Of (address): _____

_____ STATE: _____ P/CODE: _____

do hereby second the nomination by _____ for

_____ to hold the position of _____

signed: _____ Dated: _____

I, _____

Of the above address, accept nomination for the position of

signed: _____ Dated: _____