

KEEPING IN TOUCH



OCTOBER 2011

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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International: www.cdlsworld.org

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

From the Editor

It's AGM time again. Please consider nominating for the CdLSA management committee, you would be warmly welcomed. It's a great way to get a bit more involved without a huge time commitment. Meetings held by phone (costs paid by CdLSA). Details later in this KIT.

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 Association
(Australasia) Inc
PO Box 20
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PRINT POST APPROVED
PP 255003102345

6th International CdLS Conference, Copenhagen 2011

Report adapted from the UK's *Reaching Out* with thanks

Copenhagen proved to be wonderful indeed for the 6th International CdLS conference. The picturesque city offered so many attractions for families to discover. The Radisson. Blu Scandinavia also proved to be an ideal venue for this event.

Thursday 28th July was registration day and a chance for families to meet up and chat. The evening was commemorated with a reception at the City Hall. The Danish Health Minister gave a speech of welcome and there was a feast of locally-made pancakes laid on for delegates. Conference Chairman, Henrik Hasselstrom also welcomed delegates to the conference.



The pancakes await!



Australian delegates: Jenny Rollo, SAC rep. Dr Meredith Wilson, Peter Crawford and Dr Jane Law

Friday was the full Conference Day 1. Dr Angelo Selicorni introduced the first speaker. Birgit Barnes, a social worker from Roskilde Hospital, Denmark, spoke about the Danish welfare system with regard to families with handicapped children. Dr Jane Law of New South Wales, Australia talked about Health Assessment of adults with CdLS at a specialist Developmental Disability Health Unit in Sydney, which holds 2 to 4 specialist CdLS clinics per year. Next to the podium was Feliciano Ramos with an insight into Spain's Rare Diseases Strategy Made True. CdLS patients are given multi-disciplinary treatment at a specialist centre in the University Hospital Clinic in Zaragoza in Spain.

Dr Jo Moss from the Cerebra Centre for Neurodevelopmental Disorders in the UK gave delegates an insight into social and behavioural differences in people with CdLS. The address covered Autistic Spectrum Disorders as well as social anxiety and repetitive behaviours. The message was that while CdLS people MAY display ASD characteristics, there are differences and a degree of variability with CdLS.

Her colleague from Cerebra, Professor Chris Oliver gave the conference an address on the behavioural phenotype of CdLS. As well as an update on self injurious behaviour findings in CdLS, he also touched on Autism within the syndrome. Both he and Dr Moss said that Autism and/or ASD is different in CdLS – “ASD but not quite!” A diagnosis of either may not be entirely accurate but if it helps in gaining, support, information or access to benefits and services, parents should go with it, while recognising the differences. Professor Oliver also showed that many CdLS behaviours are rooted in the frontal lobe part of the brain which manages some aspects of memory and inhibits inappropriate behaviour. It is anomalies here that lead to many of the behaviours seen in CdLS. His words on Self Injurious Behaviour were that pain, particularly from reflux, is often the cause. He cited an instance where the source of pain was treated and the behaviour improved dramatically. Challenging behaviours also change with age and may get worse. Conclusions from both addresses were that environmental and social changes need to be introduced gradually so that CdLS people can cope more easily, especially as they get older.

First session of the afternoon was from Dr Helle Hjalgrim from the Danish Epilepsy Centre. Epilepsy can often be misdiagnosed or confused with other types of seizure. It is therefore important to get an accurate diagnosis confirmed as anti-epilepsy drugs can have side effects. Dr Antonie Kline from the Harvey Institute for Human Genetics in Baltimore, USA gave an address on ageing in CdLS. CdLS people age prematurely with regard to the skin and musculoskeletal system. There should be regular checks on all of the facets of CdLS as they get older. Two areas of concern were bone density, (osteoporosis) and reflux, which if untreated, can lead to Barrett’s Oesophagus and potential formation of pre-cancerous cells. Dr Marjorie Goodban from Elmshurst College USA gave a talk on Prognostic and Treatment Recommendations for Speech & Language Development in CdLS. She showed a demonstration video of an Augmentative and Alternate Communication application that runs on the iPad called PROLOQUO2GO.

The evening of the first full conference day was finished off with a Danish get together called a “Hygge.” Tea and coffee was offered with some sweets and snacks at candlelit tables while families and friends chatted together.

Day 2 began with an address by Dr Angelo Selicorni from Italy. His theme was Different Genotypes and Phenotypes of CdLS and what we have learned about them so far. One of the overriding conclusions was the wide variability of CdLS and that even those individuals with one of the known gene mutations present (about 55-60%) have very different symptoms regardless of the genetic diagnosis. Dr Anna Cereda, also from Italy dealt with the specifics of the Mild CdLS Phenotype. Next was Dr Natalie Blagowidow from the Harvey Institute in Baltimore. Her talk was on the Gynaecologic, Urologic and Sexual changes with Puberty. Most of the issues are the same as for most of the population but development is delayed overall in CdLS. There are also extra difficulties with the associated intellectual disability of CdLS.

The afternoon was concerned with gastro-intestinal issues as discussed by the next 3 speakers. Dr Peter Gillett from Royal Hospital for Sick Children, Edinburgh, UK talked about reflux as the main topic. He outlined his policy of getting on with treatment when a patient presents with symptoms of reflux rather than going for a battery of tests. If medical treatment is unsuccessful then tests are necessary. Surgery is considered only when medical therapies are unsuccessful or if there is a high occurrence of airway problems. As always, the message is very clear. Reflux must be treated because of the pain, because of behavioural issues resulting from pain and because of potential complications like strictures, Barrett's Oesophagus and airway infections.

Dr Mathilde Hansted from Hvidovre Hospital presented a Danish study into Reflux, Constipation and Urinary Tract Abnormalities in CdLS children and adolescents. This revealed some issues with correct diagnosis and treatment of reflux in the Danish Healthcare system. The last session was from Dr Margaret Marcon from Hospital for Sick Children in Toronto, Canada. She presented an ongoing pilot scheme for Constipation and Toilet Training in children with Autism or ASD. This is due to be concluded by March

No International conference would be complete without the Gala Dinner and this year's event was one of the best. Over a three course meal, delegates were also treated to a feast of entertainment. Background music was provided by the Trio La Fontaine. First of the special guests were De Kongelige Tenorer which could be translated as "The Three Tenors" – from Copenhagen's Royal Opera, no less! They were supplemented by a Soprano and gave renditions of various operatic arias – with a high degree of comedic mucking about! Later, there was the vocal talent of 13 year-old Thomas Melstrup, from Denmark's Got Talent, who brought the house down with his selection of contemporary ballads. The rest of the evening was devoted to dancing to Trio La Fontaine.



CdLS Facebook friends stop for a group photo.

The Federation of National CdLS Groups held talks over two days before the family meetings began. The eleven countries represented at this conference (out of 25 member countries) gave updates of their activities. Ulla Mugler (Italy, Chair) and Peter Crawford (Australia, Treasurer) presented reports covering the business of the Federation over the past two years. The HUB of the world group is the web site where the multi-language data base of “Ask the Doctor Questions” can be accessed thanks to Gerritjan Koekkoek from the Netherlands who so ably keeps the site updated and running smoothly. The Federation Council and the SAC work closely to ensure the latest information about treatment protocols are available to families around the world, and that families are supported even if there is no National group in their country.

Argentina was admitted to the Federation and was unanimously voted as the next host of the Federation and SAC meetings in their first National Conference to be held in 2013. The date and site of this meeting is yet to be decided. All are welcome to attend.

Elections to the executive positions were held. Chair: Ulla Mugler (Italy), Vice Chair: Henrik Hasselstrom (Denmark), Treasurer: Peter Crawford (Australia) and representing the next host nation: Paola Mannucci (Spanish-speaking countries).

Thank you to Denmark – and to Henrik Hasselstrom in particular – for hosting this excellent 2011 world meeting.



Federation Council representing CdLS families worldwide. Chris Oliver and Angelo Selicorni (Standing, 4th and 3rd from right), incoming and retiring chair of the SAC.

Welcome to new families from Singapore, WA, NSW, Vic and SA

Membership Fees paid: Rollo, Howe & Crawford Families

Donations:

General: Louise Tagliante, Marina Defina, Peter Crawford

Regular Deduction: S. Kinchela

QLD Fundraiser by Rose Humphrey

During *Disability Action Week 2011* (18-24 September) I held a small event within my workplace to raise awareness for Cornelia de Lange Syndrome. As I work for a not-for-profit organisation which supports people with a mental illness, my CEO was happy to lend his support and encouragement.

So on Wednesday, 21 September 2011, staff were asked to wear something green and/or maroon and give a gold coin. I also ran a competition to guess the number of lollies in a jar - \$1 per guess. Everyone got into the spirit of the event with staff from our Toowoomba and Maryborough offices sending down donations and guesses through the post.

I was overwhelmed and humbled by the generosity shown by my 81 work colleagues. In total we raised \$541.75 for the Cornelia de Lange Syndrome Association. This figure far surpassed my original target of \$200, thanks mainly to the very generous anonymous donation of \$200, with the message that "anything else donated would be a bonus".

The winner of the jar of lollies was very happy and generously shared them with her program at their staff meeting. I heard that the "Jaffas" were everyone's favourite.

I would like to thank my work colleagues for their generosity and my CEO for his recognition and appreciation of all not-for-profit organisations within the disability sector.

NOTICE OF ANNUAL GENERAL MEETING

The CdS Association (Australasia) Inc will be holding its Annual General Meeting on Saturday 12th November 2011 at 2.00pm Eastern Summer Time. This will be a telephone conference. Members can attend by responding with your phone number to the Secretary by end of business on Wednesday 10th November.

Only those who are paid members can vote, or nominate for positions on the management committee.

Nomination, Proxy Voting and Membership forms are included in this issue of KIT.

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: _____

**APPENDIX 1.
(ARTICLE 7, SECTION 11 (B))**

FORM OF APPOINTMENT OF PROXY

I _____
(full name)

of _____
(address)

being a member of _____
(name of incorporated association)

hereby appoint _____
(full name of proxy)

of _____
(address)

being a member of that incorporated association, as my proxy to vote for me on my behalf at the general meeting of the association (annual general meeting or special meeting, as the case may be) to be held on the _____ day of _____ 20__, and at any adjournment of that meeting.

My proxy is authorized to vote in favour of/against (delete as appropriate) the resolution/s (insert details if desired).

(signature of member appointing proxy)

(date)

NOTE: a proxy vote may not be given to a person who is not a member of the association.

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 and sent to The Secretary, P.O. Box 20 Putney, NSW, 2112, AUSTRALIA.

Credit Card:

Mastercard _____ Visa _____ (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.