

**KEEPING
IN
TOUCH**



**DECEMBER
2011**

Cornelia de Lange Syndrome Association (Australasia) Inc.

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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

Your Committee for 2011/2012 remains the same:

President: **Peter Crawford** (ACT) Vice President: **Steve Sandilands** (WA)
Secretary: **Jenny Rollo** (NSW) Treasurer: **Brett Howe** (NSW)
Ordinary Members: **Rose Humphrey** (QLD), **Lynne Wood** (VIC), **Madeleine Witham** (NSW),
Claudia Dale (VIC), **Carol Duffee** (NZ), **Lil Routley** (SA) and **Jo Argent** (NSW).

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
President's Report
2011**

This year we've fully embraced technology with our members present at the AGM almost entirely by telephone hook-up.

Once again on behalf of all our members I express our heartfelt thanks to those most involved in achieving our goals:

- our wonderful Secretary, Jenny Rollo;
- our editor of *Keeping In Touch*, Joanne Argent;
- our Treasurer, Brett Howe;
- our website manager and OZNZ on-line group administrator, Rose Humphrey;
- our state co-ordinators, Jenny in NSW, Rose in Qld, Lynne Wood in Victoria, Sharyn Burston in Tasmania, Lil Routley in SA, Steve Sandilands in WA, and Phyl Crawford in ACT; and
- our committee, Jenny, Rose, Brett, Joanne, Lynne, Steve, Lil, Madeleine Witham, Claudia Dale and Carol Duffee our New Zealand representative.

The repetition of quite a number of names in the above list shows both how dedicated some of our members are and how much is owed by so many to so few.

We abandoned our plan to bid for the 2013 International Conference to be held in Sydney. Jenny wisely decided that her involvement with RASAIID would not allow her to devote sufficient time to preparing for such a Conference. We wish RASAIID every success with their Supported Accommodation Project. Our plan for the coming year is to hold a clinic in Sydney.

I reported last year that, at the 2009 International Council meeting, it was decided that future meetings of the Council and of the International SAC should be held in conjunction with National Conferences rather than a single country putting on an International Conference. This thinking was modified somewhat by our experience in Copenhagen. We saw the greater benefit of holding our meetings in conjunction with a regional conference. So the 2013 conference will be held in Argentina but for the benefit of all the Spanish speaking countries of South and Central America.

I report that the Council and SAC meetings were fruitful. Several committees were formed to make it easier to put into effect the decisions that have been made and to plan future activity. Among these is a project to revise all the treatment protocols and express them in a way which will be helpful to parents. An information technology committee has started to revise the information in the Q&A database to bring both the USA's and the CDLSWorld's versions into harmony.

Both at home and abroad, we continue to face the challenge to achieve better outcomes for our CdLS community.

Peter Crawford
President
10 November 2011

Association News

CdLS Clinic Days for 2012 Wednesday 29th February and Wednesday 15th August.

Located at Ryde in Sydney, this is a specialist clinic for adolescents and adults, but children can also be seen.

Patients from anywhere in Australia are welcome to attend. There is nearby accommodation available for out-of-towners (at your own expense). Ask Jen for travel and accommodation advice.

Dr Jane Law takes a full medical history and gives a thorough physical examination. She refers to medical specialists and allied health professionals as needed, and writes a full report for the patient and their General Practitioner for follow-up care once you are home.

Each day there are 2 appointments for new patients, each taking 2 hours, and two follow-up appointments for one hour each. If you would like to take one of these appointments for your CdLS person please contact Jenny, or you can make an appointment directly with the clinic's Nurse Coordinator, Nicky Shellard by ringing (02)9808 9287. Nicky will send detailed instructions about what you need to bring once you have an appointment.

Perth Clinic Day

A clinic will also be held in Perth in late April 2012 if there is enough interest. Western Australian families should contact Jenny Rollo or Steve Sandilands.

Upcoming Events

Major Fundraising dinner at Mancini's (Sydney) - March 2012

National Meeting / AGM / Multi-Disciplinary Clinic - November 2012

New Families

Welcome to our new families from Tasmania, Victoria, Western Australia and South Australia.



2011 Queensland Xmas Party *by Rose Humphrey*

For the 4th year in a row, John and Margaret Wood opened their home to welcome Qld CdLS families for a sausage sizzle to celebrate Christmas.

In attendance were 4 families and they enjoyed catching up on the children's progress and achievements. For most it had been a very busy year.

Katie (20) now attends a day program a couple of days per week but keeps her mum Mary-Ellen busy the rest of the time, walking around parks, shopping centres etc – as long as she is on the move, Katie is happy.

Krystal (18) finishes her formal education this year and has some job opportunities lined up. She nagged poor mum Sue for an iPad all afternoon when she spotted Laura (14) with one.

Isabella (8) is her usual active self and now shares the attention with a 14 month old nephew Oliver. The extended Wood family have no problem though in spoiling all the children, including Justin.

Di and Richard Tetley faithfully attend every year even though their grandson Sean (8) now lives in WA. Sean is progressing well after a few medical issues recently.

Laura (14) was quite happy to entertain herself (and everyone else) with her iPad and graciously allowed Krystal to have a go – that's more than she does for us at home.



Back: Sue, Rose, Di and David

Front: Mary-Ellen, Katie, Krystal, John, Isabella (on his lap), Laura and Justin

Studies of Behaviour in CdLS

The CdLS Association encourages research participation of its member families to better understand aspects of CdLS. Recently a follow-up survey of behaviour was distributed to families who have been tracking the behavior of their son or daughter with CdLS. This study has been on-going for a number of years now, and gives not only an insight into general behavior patterns in CdLS, but a comprehensive personal report for participants. Parents have found this to be very useful for support workers to better understand their child.

There are still a few outstanding surveys. It's not too late to respond to this survey. Please post directly to the address below.

New participants in this program are also very welcome to contact Prof. Oliver and his team of researchers. This message is from Chris Stinton

The Cerebra Centre for Neurodevelopmental Disorders at the University of Birmingham (UK) and Professor Chris Oliver would like to thank all of the families who participated in their recent questionnaire study. There was an error involving dates of birth that caused some families to receive the wrong forms. The problem has now been resolved and should not affect any future mailings. The patience and understanding of all the families is much appreciated. We are currently in process of analysing all of the information and each family will soon receive a personalised feedback report. In the meantime, if anyone would like more information about the research or would like to take part please do contact us at:

Cerebra Centre for Neurodevelopmental Disorders
School of Psychology
University of Birmingham
Edgbaston B15 2TT
England

Email: cndd-enquiries@contacts.bham.ac.uk<<mailto:cndd-enquires@contacts.bham.ac.uk>>
Telephone: 0121 414 7206

Education Practices for Students with CdLS

Hello to all educators working with school-age children who have CdLS.

First, please note that the word "educator" includes teachers, parents and therapists.

The president of the CdLS World-Wide Federation, Ulla Mugler, and I would like to invite all of you to join us in looking at educational practices on behalf of students who have CdLS. We are hoping you will join us in looking at educational methods and techniques you have found helpful in your role as educator and, subsequently, begin a newsletter of *ideas*. Shortly after the Christmas holidays, we will send out a questionnaire so we will have an idea of what ideas you are finding helpful for what type of student who has CdLS. Thus, the newsletter will be your newsletter of your *ideas* which will benefit all of us.

For now, it would be helpful if you could email your name, email address, your educational or parent position and country. We welcome any and all. We look forward to hearing from you.

Ulla Mugler
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Mary Morse
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Mary Morse USA
Special Education Consultant - SAC

The following Treatment Protocols are from the CdLS UK and Ireland website, with thanks.

<http://www.cdls.org.uk/treatment/index.htm>

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.



David learning to use his new iPad. It is hoped that this will eventually be used as a communication tool.

Music therapy has been very worthwhile, and private singing lessons in the past year have helped David's speech become clearer, and use longer utterances.