

**KEEPING
IN
TOUCH**



**February
2009**

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

Correspondence to: Secretary, PO Box 20, Putney NSW 2112.
Phone (02) 9809 0287. Email: jennyrollo@bigpond.com

KIT Editor: Email: josarge@hotmail.com

Websites: Australasia: www.cdlsaus.org
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

Lots of Association news in this issue. For those who wish to attend the Melbourne CdLS clinic day on Saturday 4 April, it's important that you return the registration forms quickly. More details inside.

With best wishes to families for a good start into school and other programs,

Joanne

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.

Membership

Thank you to all those who paid their annual memberships.

Donations

Our thanks to the following individuals and organisations for their generous support:

Regular Deduction – S. Kinchela

In Kind – Riverside Girls High School for the use of their school hall for the Shirley Valentine performance, and student volunteers. Bayview Hotel Gladesville for use of wine glasses.

Computershare for printing and postage of this issue of KIT.

Staff of Mancini's restaurant donated their time at the fundraising evening.

General - NSW - Wicks, Jackson & Yapp families, L Casamento. QLD – Baillie & Wood families.

Corporate – Kimberly-Clark Australia

Shirley Valentine fundraiser – Dr F Long, J Tierney, Mr & Mrs W. O'Day, Passionist Family Group Movement, B & H Wilder, P Lubrano, A Fordham, J, Curteis, Small & Gunn Pty Ltd, M Lea, P McGee, A McGee, students of Riverside Girls High School, Mr & Mrs Hannan, Mr & Mrs Hamer, M Hicks, The Elanora Wanderers, Hempstead Marine Services Pty Ltd, S & M Homan, J Baker.

Mancini's fundraiser – *Mancini's Ristorante Italiano, Mancini's Staff, Notaras Seafoods Pty Ltd, B & D d'Apice, T & R Pinzone.* Auction donors: Mancini's, AME Products, ACN, J Rollo, LS & D Howitt, P Howitt, G Pearce, M Vie, Beth Samuel Jewellers, Strathfield Simple Technology, BP Belfield, P Rahme, Mitre 10, Iskender 12GI Optometrist, Fosters Group, JVC, Beaute Instyle, Grapevine Cellars, T Russell.

In Loving Memory of Dorothy Jean Fitzpatrick, grandmother of Jody Scheer (CdLS, from SA) – B Scheer, D & L Barrington, G Corby, H & B Osley, J Fitzpatrick, P Williams, S & C Schultz, D May, P Galliford, T Lyons, K Harris, J Cooper, B Harris, G & PBowyer, anonymous.

Welcome to new families from NSW.

CdLS CLINIC and COORDINATOR MEETING - MELBOURNE

The CdLS Association is holding a clinic day on Saturday 4th April 2009. Dr Jane Law, who runs the adult CdLS clinic at the DDHU is generously giving her time to come to Melbourne to see patients at this meeting. Dr Law will assess patients' general health and refer as needed. Please bring with you: your child's medical records folder and Medicare card.

We expect this clinic day to be popular so get in quickly to ensure your appointment. We are also negotiating with another specialist physician as we go to press.

A group dinner is being arranged for Saturday night. Costs will be extra but kept to a minimum. Details will be available on the day.

Grateful thanks to Management Committee member from Victoria, Lynne and her husband Sean, who have negotiated an excellent rate with the hotel.

What will it cost you? Accommodation (see accompanying sheet), travel and optional dinner. Doctor consultation is Medicare only.



Quality Inn Baton Rouge
1233 Stud Road
Rowville VIC 3178
Phone: (03) 9764 5050
Fax: (03) 9764 5148
www.batonrouge.com.au

(South East Melbourne)

For **accommodation** please book directly with the hotel using the form included with **KIT OR** by telephone. You must quote the CdLS Conference to get the special rates. CdLS Association will be sponsoring families who are financial members by paying registration costs. This will include lunch and am/pm teas, and any conference handouts.
Other delegates \$50 per person.

Co-Ordinators' Meeting will be held on Sunday morning 5th April following breakfast. Coordinators will be contacted shortly with a personal invitation.

Please fill in the registration form included in this KIT, and send to the Secretary. PO Box 20, PUTNEY, NSW 2112 or by email to jennyrollo@bigpond.com

NSW DEVELOPMENTAL DISABILITY HEALTH UNIT

The second half of 2008 was a time of celebration for the doctors and patients of the NSW DDHU in Putney. Spirited lobbying by the clinic's support group, with tremendous help from the then Member for Ryde and Deputy Premier, John Watkins, resulted in a funding package from the state Department of Health which guarantees three more years and increased clinic services to NSW patients with disabilities.

The support group and staff were delighted to welcome our Patron, NSW Governor Marie Bashir AC for a tour of the clinic and a celebratory afternoon tea. After formal speeches the Governor enjoyed mingling with the patients, their parents, and staff.



DDHU support group's hard working voluntary Secretary, Jill O'Connor from the NSW Down Syndrome Association and clinic patient Bridget, thanked the Governor with freesias.



NSW Governor Marie Bashir AC with the clinic's founder Dr Helen Beange AM



Local Federal MP Maxine McKew, David, DDHU lobbyist, Jenny Rollo OAM & Dr Jane Law

The following week the clinic's support group and staff welcomed Bennelong MP Hon. Maxine McKew for a tour, discussions with the doctors and an informal afternoon tea.

Patients Tania and Anna with their mums, Hazel and Helen, took part in the CdLS clinic day in October.

Dr Jane Law, who will be seeing patients at the Melbourne clinic weekend in April, holds a CdLS clinic twice a year for adults with CdLS. Initial appointments are for 2 hours when a thorough examination is given and patient history is taken. Referrals to specialists and therapists are arranged as needed. The clinic doctors work in conjunction with the patients' GP to ensure continuity of treatment. Patients with CdLS can be seen at other times by appointment, and follow-up appointments can be made at any time.

The clinic is open Mondays and Wednesdays only. For appointments call Nurse/Administrator Nicky on 9808 9287.



INTERNATIONAL CdLS CONFERENCE - Brighton, ENGLAND



The 5th international CdLS conference will be held in Brighton, England. The family conference is scheduled for Thursday July 23rd (evening) to Sunday July 26th (breakfast). Scientific and governance meetings (for SAC members and national group leaders) will be held on Wednesday July 22nd and Thursday July 23rd.

Join with CdLS families from around the world to listen to the experts' latest research findings and book private consultations with doctors from many disciplines. The highlight of any CdLS gathering is the fellowship with others who understand. World conference magnifies this aspect inestimably!

The CdLS Foundation UK and Ireland have chosen a magnificent venue for our meeting – the Metropole Hotel. They have also subsidised the accommodation and negotiated a cost per room to make it affordable for families. Never again will you have such low prices!

The CdLS Association will be sponsoring attendance of Australian financial members by paying registration costs and gala tickets. To take advantage of this significant saving for your family you will need to register before May 20th 2009.

For registration contact Jenny or for more information see the brochure on the internet at

http://www.cdlsworld.org/international/conference_registration_booklet.pdf

UK NEWS

CdLS groups from around the world have supported Prof. Chris Oliver and his team in their bid for a Centre for Neurodevelopmental Disorders to be established in the School of Psychology at the University of Birmingham. We are delighted to report that the Cerebra Centre is now open. The focus of the work of the Centre is the atypical cognition, emotion and behaviour of children and young people with genetic disorders and/or intellectual disability. You can find more information on the activities of the centre on their website (www.cndd.bham.ac.uk)



🎄 CHRISTMAS IN THE PARK 🎄

Santa came early for Qld CdLS families when he made an appearance at the Christmas Picnic in the Park on Sunday, 30 November 2008.

Twenty-six people – 5 (CdLS), 15 adults and 6 kids – attended the fully catered picnic at their usual location at Orleigh Park on the banks of the Brisbane River. Parents caught up over drinks and nibbles while Maddie Humphrey and her friend Georgia entertained the kids with face painting and other games. The men took on the cooking duties for the lunch sausage sizzle, while the ladies continued to catch up. Dessert of chocolate mud cake doubled as a birthday cake for Katie (CdLS) who would celebrate her 18th birthday on Christmas Eve.

After lunch Santa and Mrs Claus arrived with sacks of gifts for the children and adults alike. Every child received a huge bag filled with goodies ... stuffed toys, beach towels, sports caps, bubbles, lollies, just to name a few; and adults received mugs, caps and T-Shirts compliments of AMC Cinemas.

Many thanks go to our sponsors – IGA McDowall for nibbles and soft drinks; McDowall Quality Meats & Chicken for the sausages; Brumby's McDowall for bread rolls and Woolworths Brookside for a ¼ slab decorated chocolate mud cake. Many thanks also to Margaret Wood who organised the donation of goodies and packing of Christmas gifts for everyone.



Santa with Kaden, Laura, Sean, Isabella & Kate
(Parents give Santa a helping hand!)



Parents admiring their gifts of AMC
T-shirts, caps and mugs



Laura, Sean and Isabella with Santa

Fundraiser - *Mancini's Ristorante Italiano*, Belfield, Sydney.

The owners of *Mancini's*, Eric and Gordana Mancini, wanted to contribute to the community by holding a fundraising dinner for a local charity. The CdLS Association was chosen by one of their staff managers, Sandra, who has a personal association with CdLS.

Over 140 guests attended, including many of the auction item donors, member for Canterbury and Minister for Community Services Hon. Linda Burney MP, and celebrity entertainer & fundraiser Maria Venuti AM, who sold the raffle tickets in her inimitable way! CdLS families were well represented by the Rollos (David aged 25yrs), Hallings (Sean aged 17yrs), Withams (Ella aged 13yrs), and Sandra whose daughter Daniella passed away in 2002 at the age of 7.



Part of the 140-strong crowd who came to support CdLS families.



Special guest, Maria Venuti AM with *Mancini's* manager Barry Leslie, who was MC for the evening.

The restaurant donated 100% of the food takings to the association. Staff donated their time working on the night, sought out many of the auction items, and took up a collection as well. Gabi, Kerry and Barry were crucial in the organising, and Barry acted as MC. Auction items included some stunning jewellery, wines, an autographed football, car accessories, spa treatment, autographed CDs and posters. Many businesses local to the restaurant became involved and were very generous in both donating prizes and bringing people along on the night.

Everyone really enjoyed themselves, the food was great, the staff very friendly and efficient, and lots of money was raised. And did I mention the food was fantastic?!

Mancini's have kindly offered to do it all again this year. We are therefore requesting donations of items suitable for silent auction and raffles. Can **YOU** help? And do come along and join in the fun! Details will be published in KIT later in the year.

TREATMENT PROTOCOLS Thanks to CdLSUK web site



Hearing

People with CdLS may have very tiny structures and testing may be difficult. It is advisable to consult an audiologist and/or otolaryngologist who is familiar with CdLS or who is experienced in working with infants. Pharyngeal-oesophageal tubes may be useful for middle-ear drainage as needed but a physician experienced in working with small infants is usually necessary.

If a hearing loss is suspected, headsets and hearing aids should be prescribed for infants and children. Even a mild hearing loss can result in a speech and language delay. Smaller aids are available so it is not necessary or advisable to use an adult-sized aid. If the child will not leave on the aid, an audiologist or behavioural therapist may be helpful. Appropriate audiological management should include selection and fitting of suitable amplification for all listening environments. While the child's personal hearing aid may be sufficient some of the time, the use of FM amplification may be necessary in other situations.

Almost all children with CdLS are diagnosed with mild to moderate and sometimes severe hearing loss, however interviews with caretakers reveal unexpected reports regarding the history of audiological results. Many caretakers report that their children were diagnosed as severely hearing impaired at birth, moderately impaired at 12 months, and mildly impaired or without impairment at age 2 years. Since it is unusual for hearing to improve rather than worsen, it seems correct audiological assessment is difficult. Individuals with CdLS may have narrow ear canals and difficult behaviours, making examinations a challenge.

Parents often report their children seem to hear much better than their test results would indicate. Considerable confusion exists regarding hearing ability for some children. Many children fitted with hearing aids will not tolerate the use of them or do so only sporadically. For these individuals, retesting is often important to insure that the aid is beneficial. There have also been reports of young children fitted with adult-sized hearing aids when child-sized aids are available and more appropriate.



Dental

Dental problems can include small jaw development, poor oral hygiene, crowded teeth, small teeth, periodontal disease, and the erosion of teeth caused by stomach acids from reflux. Children diagnosed with CdLS require early dental evaluation, treatment, planning and preventative home care. The oral structures are greatly affected with this syndrome. Growth and development concerns of the jaws and teeth must be assessed at a young age in order that therapeutic measures can be instituted.

Clefting of the palate is common in patients with CdLS. This condition would necessitate pediatric dental involvement shortly after birth. Feeding obturators and surgical stents may be of great assistance to the craniofacial surgeons. The early introduction to dental care allows for baseline documentation of the oral soft and hard tissues. Clefts should be repaired as early as possible. Not only does this repair improve the ability to eat and reduce the likelihood of ear infections leading to hearing loss; it also enhances the speaking process.

Oral hygiene practices can be taught to caregivers, thus, preventing infant bottle cavities and other common dental problems. Routine six-month check-ups can assist in monitoring changes, detecting pathology and reinforcing appropriate home care. Orthodontic evaluation should take place by the 7th year.

Pediatric dental specialists are trained in the specific dental requirements and treatment of children, especially in the area of children with special health care needs. The American Academy of Pediatric Dentistry recommends regular professional attention beginning with patient oral evaluation and parental counseling no later than 12 months of age with a pediatric dentist.

Pediatric dentistry educators and practitioners throughout the country are advocating a preventative approach, which has been termed the no-cavity plan. In addition to water fluoridation, optimal systemic supplemental and topical fluoride, the daily use of appropriate amounts of a fluoride dentifrice and the placement of pit and fissure sealant, the no-cavity approach utilizes nutritional counseling, strict home oral hygiene care, the maintenance of existing restorations and biannual professional cleanings.

When indicated, using conscious sedation and performing a number of tasks at one time may optimize dental care as well as care for other medical conditions for certain individuals. Individuals who are unable to communicate pain or discomfort and who are uncooperative in the dental office may need to be sedated in order to provide quality care. Remember, whenever an individual has anesthesia for dental care, consider other procedures that could be done concurrently (endoscopy, ENT exam, eye exam etc).

Jenny Rollo has Collis curve toothbrushes for sale in adult and child sizes for \$5 for financial members. The cost to others is \$10 to pay for postage and packaging.

CdLS Clinic MELBOURNE

REGISTRATION FORM

Name: _____

Address: _____

Telephone: _____

Email: _____

Saturday 4th April – Clinic Day

Number of ADULTS attending - _____

Names: _____

Number of CHILDREN attending - _____

Names: _____

PERSON/S WITH CdLS - _____ **Age/s:** _____

Name/s: _____

I need an appointment with Dr Law for:
initial assessment _____ review _____

Sunday 5th April – Co-Ordinators' Meeting (by invitation)

9am to 11.30am Yes I am attending this session.

Name: _____

Have you booked your accommodation?
Not a financial member of the CdLS Association?
Number attending _____ **X \$50 =** _____
Please fill in banking details on the membership form.

Send to the Secretary. PO Box 20, PUTNEY, NSW 2112 or by email to jennyrollo@bigpond.com