

**KEEPING
IN
TOUCH**



**December
2005**

Cornelia de Lange Syndrome Association (Australasia) Inc.

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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

I hope KIT finds you relaxing, feet up, having a few days off over Christmas enjoying family and friends, with time to read in peace. (...we can all dream ...)
A happy and healthy 2006 to you all. Cheers!

New Members

Welcome to our new members from Iran, Victoria and New South Wales

Sponsor of our Web Site :

Would you like to be the web sponsor for a year, or part of a year? Please contact the Secretary. You, too, can have your name in « lights » on our web page ! If you haven't visited the site for a while, check out the changes and additions Rose has made.

ELECTRONIC KIT NOW AVAILABLE !

If you would prefer to receive your CdLS Newsletter in electronic form by email in PDF, please let us know. Just email Jenny (address front page) with ELECTRONIC KIT as the subject, and – voila !

AUSTRALASIAN CdLS miniCONFERENCE

Your CdLS Management Committee is delighted to announce that a mini conference will be held in 2006. We hope to finalise plans soon, so that full details will be available in the next issue of KIT. In the mean time, save the weekend of **30th September/1st October 2006**, and prepare to travel to the glorious **Gold Coast, Queensland**. We expect to dedicate the Saturday to meetings and workshops, and the Sunday to more social pursuits. Looking forward to seeing you there.

COMMONWEALTH GAMES QUEEN'S BATON RELAY

Keep an eye out during news reports on January 25th – the first day of the Australian leg of the Queen's Baton relay in Australia. Our Australian CdLS families will be represented by our intrepid Secretary. Jenny will be taking her turn with the Baton for a short 500m stint in St Mary's, a western suburb of Sydney. (If the baggy shorts don't look TOO daggy, we'll print a photo in next KIT !!)

MEDICAL RECORDS FOLDERS

In association with the NSW Developmental Disability Health Unit and the Centre for Developmental Disability Health Victoria, the CdLS Association has developed a medical health record folder for its members. One will be sent to each family by February. In it you will be able to record details of your child's medical procedures, illnesses, vaccinations, weight and height records, medications, seizure activity, etc, etc.

These records are ideally to be used from birth, but it's never too late to begin using it. The folder is intended to be a permanent record which stays with the person through their life and residential moves. This will ensure that any medical practitioner who sees your child will be immediately able to understand CdLS, and have a clear medical history of the patient. No longer will parents struggle with remembering dates of medical events!

COLLIS CURVE TOOTHBRUSHES

We still have some collis curve toothbrushes available. Please contact Jenny .

AGM and PICNIC 2005

The AGM was held on the 29th of October, at Casa Rollo. Jenny, the Argents, Steve and Peter attended in person, with a raft of others via the magic of the telephone conference. The election process (the highlight of the AGM), was conducted without too much money changing hands and just enough intrigue to make it interesting. 8-) The meeting was followed by the now traditional family picnic at Putney Park, where we were joined by the Halling family, and we ate and drank and watched the boats on the river until it was time to go home.

Ian

YOUR NEW COMMITTEE FOR 2005/2006

President : Peter Crawford (ACT) **Vice President :** Steve Sandilands (WA)

Treasurer : Brett Howe (NSW) **Secretary :** Jenny Rollo (NSW)

Ordinary Members : Rose Humphrey (Qld), Carol Duffee (NZ), Claudia Dale (Vic), Joanne Argent (NSW) and Kate Coffey (Qld).

We extend a very warm welcome to Kate who has joined the committee for the first time, and a warm welcome back to our very busy Editor, Joanne.

Thanks to Joe Yatras and Garry Saunders who are taking a break from committee duties. They both served your Association well in the previous year.

Cornelia de Lange Syndrome Association (Australasia) Inc President's Report 2005

How does one judge the health, the efficiency, the well-being of an association?

On the one hand, our affairs are running smoothly, thanks to the unflagging efforts of many:

- Our state co-ordinators, the quiet, behind-the-scenes workers of our outfit deservedly merit our thanks for continuing to be there for our families with that invaluable personal support and information.
- *Keeping in Touch* continues to provide that important link for our members -which highlights what a large debt we owe to its editor, Joanne.
- Our indefatigable Secretary, Jenny, is an inexhaustible source of inspiration, enthusiasm and ideas.
- The Committee strives to further the work of the Association. Thanks are owed to them for their continuing dedication and for their self discipline and efficiency in making our meetings by teleconference such a success.
- Rose has further developed our national website to its present excellent standard: www.cdlsaus.org . Thank you, Rose.
- Our bank balance is healthy! – And is well looked after by Brett!
- The idea of CdLS clinic days is spreading.
- The OZNZ on-line group continues to support its participants.

On the other hand, questions arise:

- Can we do more to identify more people who have CdLS?
- Can we increase our membership?
- Do we have throngs of people eager to take on leadership roles on the committee?
- As a small organisation, can we access government/community help in reaching out to present and future members?
- Are there better ways to bring our members together for that reviving, exhilarating, comforting experience of mutual support?

On the international level I can report:

- The Italian CdLS Association hosted a very successful, vibrant conference in Grosseto in June. Four Australian families attended and *Keeping in Touch* has provided coverage of what was described as “warm and welcoming, boisterous and animated, emotional and exciting, memorable and unforgettable”.
- Jenny and I attended the meeting of the Council of the International Federation of CdLS National Support Groups. We were delighted that Ulla Mugler, the moving force behind the Grosseto Conference, was elected President of the Council. She will bring that same drive to furthering the aims of the Federation. As I reported in *Keeping in Touch* the Council identified two major issues – language and the database of answers to Ask the Doctor questions – which the Council will be addressing over the next two years.
- On the way home from the Conference, my wife, Phyl, and I visited Auckland to meet with some of our New Zealand families to tell them about Grosseto. The Kiwis organised a mini-conference and it was noticeable what a great spirit they have fostered among their members: our thanks to Liz and Vernon Molloy for hosting us and to the whole group for the warmth of their welcome.
- Two DVDs have been produced about CdLS: one by the UK which is a moving portrayal of the ups and downs of life with a CdLS person; the other by the US which concentrates on helping people to identify CdLS persons within the community.

At our Association level I note:

- The federal government has accepted CdLS as a ‘recognised disability’ for the Carer Allowance (Child). This will be of benefit for future claimants but also for the review of present ones.
- NSW had its Third CdLS Clinic Day in September and similar events are being developed in some other States.
- A number of our NSW members worked very hard to have funding for Post-School Programs reinstated.
- The Siblings Conference in Adelaide late last year had significant CdLS input with Bernadette (the CdLS President from Canada) speaking and Nicki, Jenny and Rose contributing.
- Our Philippines sub-group held a family gathering and seminar in August.
- Our New Zealand members held a mini-conference in July.

While we can look back on a very exciting year, the challenge is to convert what we have experienced into better outcomes for our CdLS community.

Peter Crawford
President

TREATMENT PROTOCOLS

Thanks to the CdLS Foundation UK & Ireland web site.

The Role of Ultrasound in the Prenatal Diagnosis of CdLS

For parents who already have a child with Cornelia de Lange Syndrome (CdLS) and who want to have more children, prenatal ultrasound can provide a helpful evaluation of subsequent pregnancies. Research continues to support the low recurrence risk for CdLS and ultrasound can be a source of reassurance and comfort to some parents.

Ultrasound is currently our best - and possibly our only - tool for prenatal diagnosis. There are a number of findings in CdLS that can be recognized prenatally. Probably the most reliable indicator is poor growth in a small fetus. Poor growth can be detected at ages greater than 25 weeks of gestation for most babies with CdLS, especially if there has been accurate dating with an earlier ultrasound.

For parents who have had a child with CdLS, an initial ultrasound examination may be recommended for all subsequent pregnancies at 18 weeks. The timing of this scan will allow a thorough study of the anatomy of the fetus, and also provide accurate dating of the pregnancy. In addition to the routine survey, the anatomy that should be examined particularly carefully includes the face, the hands, the heart, the arms, and the ventricles of the head. A second ultrasound study at 30 weeks could provide added reassurance that the fetus has grown normally, if needed.

An initial scan at 8 weeks would provide a very accurate estimate of the fetus' age, and this would allow a close monitoring of the growth. In addition a scan would also have to be performed at 18 weeks because the organs of the fetus are not completely formed and fetal anatomy cannot be seen clearly at eight weeks.

Anatomic abnormalities that have been described on prenatal studies of babies with CdLS include: limb abnormalities (particularly of the upper limbs), abnormal hearts, cleft lip, abnormal facial profile, diaphragmatic hernia, mild enlargement of the ventricles of the head, and gastrointestinal abnormalities.

The most distinctive of these abnormalities are those of the upper limb. Arm bones and fingers should be carefully examined and counted because these may be missing or abnormally short. The femur, feet, and arm bones should be measured to ensure that their lengths are within the normal range. There are tables of normal measurements that the person performing the ultrasound study should have.

In the face of a baby with CdLS, one might find cleft lip or palate, long eyelashes, a small chin, and a small upturned nose. In many of the children, there are abnormalities of the heart (ventricular septal defects, atrial septal defects, hypoplastic aorta, persistent left superior vena cava, and tetralogy of Fallot).

The head of the baby with CdLS will tend to be short and small (microbrachycephaly) and so should be measured and compared to tables of non-normal measurements. The person performing the ultrasound study should also check to be certain that the baby does not have a diaphragmatic hernia or abdominal calcification (meconium peritonitis).

In many cases, babies with CdLS move much less than normal babies. The person doing the scan could perform a biophysical profile to provide an evaluation of the babies behaviour and responses.

Ultrasound is not a perfect tool to diagnose CdLS, nor is it a perfect tool to exclude the possibility of CdLS. Nonetheless, most children with CdLS have fairly severe abnormalities that can be detected prenatally by careful radiologists or obstetricians. It is anticipated that in almost all cases, scans performed on subsequent pregnancies will be normal. This should help parents find considerable joy and hope with these pregnancies, rather than anxiety and fear.

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.



BEHAVIOURAL OPTOMETRY

Claire Alexander

Fellow of Aust. College of Behavioural Optometry
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What is the difference between looking at a picture of an owl in a book and actually observing one in real life? That is like the difference between sight and **vision**. A behavioural optometrist sees **vision** as a part of the whole person, a part that influences how you act, react and move. We use vision to help us balance, walk, drive, play sport, talk to people, work and read.

The most common idea people have of an optometrist is someone who asks you to read the chart at the end of the room or checks your sight. This is but a tiny fraction of how a behavioural optometrist assesses vision. Most people also think that vision can not be tested without the person being able to answer questions. Vision can be tested at any age from new born on and the information we get can be objectively measured and also inferred from changes in behaviour. This means if we can get someone's eyes to stay open we can get measurements and information. The leading cause of blindness in the world today is uncorrected focus problems and these are easily assessed objectively, with no verbal response necessary. Even in Australia there are approximately 13% of people with uncorrected vision problems.

Everyone should have a vision assessment every two years. Vision changes over time and how we see at 5 years of age is very different to how we see at 60years of age. People

with special needs often have a higher incidence of vision and spatial awareness problems. An assessment by a behavioural optometrist may offer some solutions that can improve performance and quality of life. The tools we use include lenses, prisms, vision training and light therapy (syntonics).

We can use these tools to help people and change their interactions in the world in many different ways. Lenses, prisms and light therapy may make things clearer but they may also change eye contact, posture, attention and comfort. Many times we test children who are unresponsive to their environment and have never had a vision assessment. By giving children more comfortable vision they begin to interact more with their environment and communicate better.

An example of how changes to vision can make a difference can be seen from the following case.

Greg (not real name) came in because when he walked he favoured his right leg and he veered left into walls and furniture at times. Greg is 15 years of age and has cerebral palsy. He did not like reading any small print and he didn't look directly at people when they spoke to him but appeared to look past them.

Testing found Greg had a left visual field loss especially when looking down and he had a focus problem when looking at reading distance. His vision if tested on a chart in the distance is normal. We used prism glasses for general wear that brought Greg's remaining vision more central and helped him to keep better eye contact. We used lenses to help him focus at near for reading. Greg did vision therapy to teach him to use his eyes to quickly scan the left hand side and down when he was walking, which along with the prism glasses, helped him to veer to the left less. Greg now has fewer bruises on the left side of his body as he doesn't bump into as many things.

These changes helped Greg feel more confident to walk through shops and talk to people.

Once vision is comfortable we can begin to enhance performance through vision training. Vision is a learnt skill and sometimes due to physical difficulties or avoidance these skills do not develop effectively. This includes being able to keep your eyes on a thing long enough to work out what it is and where it is. We also need to learn to move our eyes from one thing to another at will. This and many other skills are able to be trained if they haven't developed and can help in improving communication and education.

To find an optometrist who has a post graduate fellowship in behavioural optometry visit www.acbo.org.au

Don't forget to consider vision as part of the whole healthcare package.

VIBRATIONAL KINESIOLOGY

by Sheila Kennedy



Vibrational Kinesiology is the term used for a simple and effective way of correcting the energy fields of the body, its aim being the optimum health of the person being treated. It does not require any manipulation, drugs or invasive procedures, and generally needs only one initial two hour appointment with one or occasionally two follow up treatments about six weeks later. Any further or maintenance visits similar to tuning your motor vehicle are at the clients need and discretion. So how does it work? The human body is built on three electrical systems, and these can be likened to the power box on the outside of your home. In your power box there is a bank of fuses or electrical switches, a bundle of wires going up into the roof and down to the lights, power points etc and power coming into the power box from whoever you buy your power from. They are known as the Chakra, Meridian, and Auric field systems.

If these three energy systems are not communicating with one another then the body cannot function correctly. Just as if you have pulled a fuse out of the fuse box there will be no power to a particular part of the house, or if the power company turns off the power you will need to use alternative lighting etc eg gas lamps, torches or candles.

In the 2 to 2.5 hour session which can be an individual or a whole family, I identify the areas that need to be corrected by using basic kinesiology which is muscle testing, and then correct the body by using ancient knowledge comprising of sound, scent and crystals. If your body requires it I will supply a free CD of sounds to be played at home to fine tune the correction.

Further information can be sourced from The Jessie Carran Centre, @ Austherapy .com, Sheila Kennedy @ Find a Therapist .com.au, or Sheila Kennedy Holistic Practitioner, or by emailing or calling me. Email sheila@jccentre.com.au, Ph 03 98524472. I am also registered with The International Institute of Complementary Therapists, as a Holistic Practitioner.

Sheila has very generously offered free treatment to people with CdLS. Her practice is in Victoria.

CdLS CLINIC DAY - NSW



Delighted to see each other, Christina and David take their turn as patients of the NSW Developmental Disability Health Unit (Formerly CDDS) CdLS Clinic Day in September.

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.
