

**KEEPING
IN
TOUCH**



**September
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

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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

Membership Fees Paid

WA - Sandilands family, Qld – Corcoran, Baillie, Dolinski families, NSW – Rollo family,
SA – Scheer, Routley families, Vic – Eisele, Troiani families, ACT - Sheila Kenny

Donations

Heather & Gordon Lee, Gerdina & Cedric Bryant, Annette Fordham, M. Nuttall & Crawford,
Sandilands, Corcoran, Baillie & Routley families

Regular Deduction

S. Kinchela

In memory of Dorothy Fitzpatrick

Judy Scheer

**Welcome to new families from NSW
Membership fees for 2009/2010 due**

CoOrdinator News

Victoria

A very warm welcome to Lynne Wood (Corey's mum) who has taken on the role of
Victorian Coordinator. Lynne is a very busy working mum to three very active boys who still

finds the time to be a member of the CdLS Management Committee. She hopes to be able to contact as many families as possible over the coming year. Victorian families are welcome to contact Lynne at the address on the front page of KIT.

We are not exactly saying goodbye to Jaci Wiley, our Victorian Coordinator of many years' standing. Jaci has been unable to give the time she wanted to the CdLS group due to work, family and studies so she is passing the baton to Lynne with her best wishes, and the knowledge that Lynne is up for the task. Jaci is still available as a trained counsellor and looks forward to catching up with families at future gatherings.

From the Committee and Victorian families, grateful thanks to both Jaci and Lynne.

Western Australia

Steve Sandilands, who has been a committee member since the beginning, and has attended many national & international CdLS conferences, has taken on the extra role of contact person for WA in place of his wife, Yvonne. Thanks to Yvonne for being the contact person for many years.

Australian Capital Territory

You may notice we have added a contact address for the ACT. Phyl and Peter Crawford have been performing this role for years in ACT and southern NSW. As the number of families in this area has increased, and we are still coordinator-less in NSW (anybody interested?), we are now formalising their duties! Thank you to both Peter and Phyl who are such a strong and reliable support to many families.

New Flyer and poster Boy

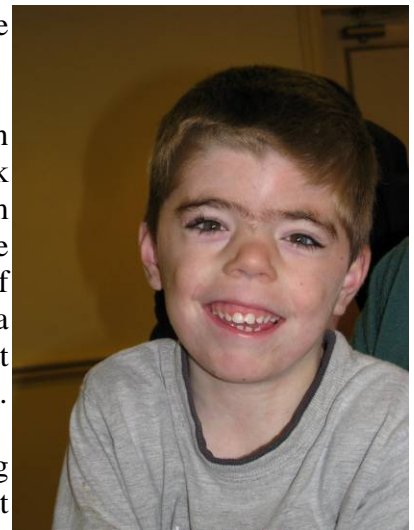
Thanks to Sarah Allen for helping in the layout of our new flyer. It was entirely coincidental that her beautiful son, Ben is the front page poster boy! – Jen

Meet Ben, our little boy who has CdLS, but loves the hell out of life anyway.

When Ben was diagnosed with CdLS, the amount of information and opinion thrown at us was overwhelming. Not much of it stuck with us, apart from a feeling that no one expected much from him and that we shouldn't either. Yes, life was really rough on everyone for the first 4 or 5 years of Ben's life, constantly in and out of hospital, all sorts of health problems, learning to cope with the idea that your child is 'different'. In this kind of a situation, it's no great surprise that you find yourself joining the naysayers to some degree.

However, it's fast approaching Ben's 7th birthday and he is defying all of the predictions made by the swarm of well-meaning, but ultimately disheartening experts that turned up around diagnosis time. Ben will always be different from the other kids, but will be by no means helpless.

Ben is now at a mainstream primary school with the help of a full time aide, enjoying grade 1 after having successfully negotiated Prep and, by all accounts, loves school and would be lost without it - he's already one up on his Dad! I had reservations about Ben attending a



mainstream school, because casting my mind back to when I went to school I could not remember being around any disabled kids - they went somewhere else, don't know where, just Somewhere Else - but it turns out I was wrong. He's made heaps of friends and has a ball at school.

There's been remarkable progress made by Ben during his time so far at school. For all the gloomy predictions, it's awesome to see him able to look at the numbers between 1 to 5 and hold up the appropriate number of fingers and have a go at saying it as well. Just recently, it's become even more so now that he's able to read his own name and a small number of words. Story time has now become a joint effort, especially as far as cars are concerned - the dirty looks you get if you don't point at the word 'car' and let Ben say the word instead of you reading it. He's also nearly mastered writing his own name, too.

Even better than the scholastic development is the emergence of a little boy with a personality all of his own. There can be moments, but in the majority he is the happiest kid going. He has a love of life and all of the people around him that you don't see very often. Nothing warms you up more than Ben charging at you for a big 'hello' hug, complete with hearty slap on the back. He definitely does not lack in cheekiness, he's successfully used the "Look behind you, isn't that Elvis?" ruse to steal Mum's last bite of pretzel from under her nose, and none shall disturb Lord Muck as he sits in his royal bed (read: Mum and Dad's bed after he's kicked them out) early on a Sunday morning, drinking his juice. A hearty backslap commiserating with Dad after handing him another defeat in Wii bowling is cheerfully given, too.

The 'no' phase is in full swing at the moment, as happens with all kids.

"Ready to go to bed, Ben?" "Nooooooooo".

"Want some dinner, Ben?" <spoken around mouthful of food> "Nooooooooo".

Hopefully the tantrum stage gets skipped altogether.

Ben loves helping around the house, too. Ask him to set the table, and he will. There'll be a plate for everybody in the appropriate place and for reasons known only to him, 4 to 6 forks for each person but no knife. That part still needs some work.

Ben has had a very rough start to life, but is making the most of it - and showing us that 'the experts' are still only people like you and I and they get things wrong occasionally, too.

By Ward Allen (Ben's Dad)

June 2009

Melbourne Clinic Day

The CdLS clinic day held in April was attended by a small number of local families. Delegates were fascinated to learn more about gastrointestinal disorders commonly affecting people with the syndrome during the presentation by Dr Barry Lipschitz, a well respected paediatric gastroenterologist from Melbourne. Barry has treated a number of patients with CdLS.

Dr Jane Law, our wonderful disability specialist from Sydney, and Dr Lipschitz saw patients individually for consultations. Parents have since been able to follow up with their local doctors, and were directed to the specialists needed for current health problems.

We kept formalities to a minimum to allow parents to talk, but of course there is never enough time for talk when our families get together!

We made the most of our gathering by conducting a very rare face-to-face committee meeting as well as coordinator training and future events planning. Being in the same room for these meetings is invaluable. As wonderful as technology is in allowing us to cheaply and conveniently meet regularly via teleconferencing, there is really nothing as productive as being together in person.

Keep an eye on KIT for future events announcements.

Dr Jane Law's motel room became her consultation room - here with Sean, Corey and new VIC coordinator Lynne Wood



Darren, Ella and Lil Routley (SA) consult with Dr Barry Lipschitz

Sheila Kenny (ACT) meets Joe, and Mary Troiani and their daughter Rosemary (Vic) who very quickly became a favourite!



Conference in Brighton, England



The families from the UK, Europe and around the world who were able to attend the conference in Brighton were not only able to get the best medical help available anywhere, but had a fantastic time socially as well! These conferences are really like a family reunion, even if this is the first time of meeting. Conversations occur spontaneously and animatedly as mums and dads, siblings and grandparents all quickly find common ground.

Doctor consultations were available with experts from many disciplines. Word quickly spread among parents about how helpful consulting with our Dr Jane Law was, and her “dance card” was quickly filled. Many people told me how lucky we are to have Jane supporting our families in Australia. (I already knew that!) Rose Humphrey helped English and Italian speaking families to make their appointments, and Phyl Crawford joined Frank Mairano (USA) to keep the appointments to time. Not an easy task!

Presentations were made by many of the experts, allowing ample time for questions and discussions. Some of these will be published in future issues of KIT.

We look forward to the next international gathering in 2011 hosted by Denmark.



We were delighted to meet Ben Allen's English grandparents, Margaret and Stephen, seen here with Peter Crawford



Some of the UK group's organising committee at the SAC & Federation welcome function at the aquarium.



Scientific Advisory Council (SAC)

- Abstracts from this UK conference will be published in the American Journal of Medical Genetics. This will encourage further research as well as offer new information about CdLS and raise awareness. Not all papers will have formal conclusions.
- Today there are new presentations and updating research from Italy, USA and Poland; and from the UK neuropsychology and behaviour
- Professor Chris Oliver (UK) has been elected Chair of the SAC effective from 2011.
- There is a lot of interest in research in genetics and practical management methods
- Australia's usual SAC representative, Dr Meredith Wilson, was unable to attend this year. Australia was ably represented by Dr Jane Law who specialises in adult disability medicine.



Emanuelle Bassile (Italy), Jane Law (AUS),
David Fitzpatrick (UK)
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