

Community Spirit



CdLS For Supercheap Auto

Most parents will admit that, at times, their feelings toward their offspring and the whole parenthood trap fluctuate like Melbourne's weather. Beautiful one day, less than perfect the next! However, there's one small group of parents who are inspired by their kids everyday and I'm fortunate to be one of those parents.

My youngest daughter was born by emergency caesarean on 25 March 1997. She weighed 5lbs, had a heart murmur, needed resuscitation and nasal-gastric feeding. At 4 months she was admitted to ICU with a respiratory virus and grand mal seizures.

At 9 months an ABR confirmed her partial deafness, and at 17 months her condition was finally diagnosed as Cornelia de Lange Syndrome (CdLS) – a rare condition, not often seen.

At diagnosis we were told that Laura would “probably not do much” however, during her short 10 years she has reached most childhood milestones, participated in mainstream education, learnt to communicate without words, smacked John

Howard's face, and provided essential data in the search for the cause of CdLS, its characteristics and methods of improving life for those born with this syndrome.

Most doctors may never see a person with CdLS in their career, so until a few years ago information was limited, outdated, and frightening. In 2001 parents, representing CdLS support groups from a number of countries, came together to form an international forum to share information and created “CdLS-World”, an international hub for worldwide organisations united by CdLS. Also created was the Scientific Advisory Committee (SAC) – a group of interested doctors who are prepared to commit their time and expertise to assist the cause of people with CdLS.

Through extensive and ongoing research the SAC have to date identified three responsible genes; a number of common medical issues have been addressed and challenging behaviour understood. This research can only be conducted using those affected by CdLS and Laura has been one of those candidates. SAC findings are presented at International Conferences held every 2 years in various host countries – USA – 2001; Australia – 2003; Italy – 2005; and Canada – July 2007.

So far I have attended each Conference as a parent. However, this year I have been invited to attend the Canadian Conference as a representative of CdLS Australia, a support group for its 150 members. In Canada I will participate in meetings with members of CdLS World to discuss our shared mission and plan the future direction of the group; and meetings with the SAC to discuss current and future research projects.

Needless to say I am honoured by this responsibility; however, Laura is the true hero who should be congratulated. Over the years, whilst struggling to belong in a world that sees her as “less than” other children, Laura has allowed herself to be examined, x-rayed, scanned, blood-tested, measured and studied, always with a mischievous grin and patience beyond her years. Even though there can be some really tough days caring for her, these do not diminish my pride in her courage, persistence and tenacity.

For more information on CdLS
visit www.cdlsaus.org or www.cdlsworld.org

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