

CONNECTIVE ISSUES

March 2008

Welcome to our next edition of *Connective Issues*, providing you with information about ConnecteD's fundraising events and any updates/news from the Clinic.

Report from Professor David Sillence

ConnecteD should be proud of its contribution over 15 years to the development of multidisciplinary services and research for children with Connective Tissue Dysplasias. One of the clinic's and ConnecteD's most important goals has been to ensure that all children have an opportunity to have a correct diagnosis, genetic counselling, multidisciplinary therapy consultation and planning and medical therapy where available. ConnecteD represents and advocates for groups of children with Marfan Syndrome, Arthrogyrosis, Skeletal Dysplasias, Mucopolysaccharidoses, Osteogenesis Imperfecta, Ehlers Danlos Syndrome *and related disorders*.

There is very little change of government funded co-ordinated care for such rare conditions in the present health care climate. So it is vitally important that our groups work so well together, as collectively we can do so much not only for individual patients and their parents but for the whole group.

Staff Activities

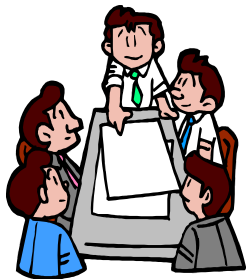
Dr Lanie Alcausin, Fellow in medical Genetics (Bone and Mineral Disorders) passed her examinations in children's bone densitometry.

Dr Alcausin and Professor Sillence will be presenting a poster on the work of the Connective Tissue Dysplasia clinic at the 7th Asia Human Genome Organisation in Cebu in April 2008.

Report cont.

Dr Alcausin and Dr Craig Munns have commenced a research study to examine whether twice daily standing on a Vibrating foot plate can enhance bone density. The study is mainly directed at improving density in children with Osteogenesis Imperfecta, however we have had children with other disorders such as Ehlers-Danlos who are being treated as an extension of this study.

The CTD service has supported the organisation of a meeting "Therapy and its Evaluation in Musculoskeletal Disorders of Childhood" on Friday 9th May, 2008. Our guest speaker will be Associate Professor Raoul Englebert, Director of the National and International School for Physiotherapy, Amsterdam, who will be coming to Australia as the guest of OI Australia for their Annual Conference, 10th-11th May, 2008.



ConnecteD Meetings

Our meetings are held approximately once a month at 7.30pm at the Children's Hospital, Westmead. If you would like to attend please contact either Jennifer Reynolds (Secretary) on 9845-6444 or Lynne Foxall (President) on 9845-3179.



Christmas Stall

Our annual Christmas Stall was held on Friday 7th December 2007 at the Children's Hospital. There were lots of bargains to be had and sales were brisk. Thanks to those Committee members who helped out on the day - Lynne, Akiko, Dr. Jenny Ault, Lauren, Gail and Marilyn. Thanks also to member Jan Schroder for those very yummy cakes that she spent all week baking and to her daughter Janelle (Marfan syndrome) for helping out on the day. The total amount raised was \$1,267.85.

Committee Members - Profiles



Dear Members and Friends,

The *ConnecTeD* Committee would like to establish this section of the newsletter, hopefully as a "regular edition". We trust that knowing a little about who is on the committee, who are members of the CTD Medical Team and members of the *ConnecTeD* Association will bring cohesion to our group.

Committee Member - Non Executive Professional Member Dr. Jenny Ault

These days I work as a senior medical specialist in the Connective Tissue Dysplasia clinic at the Children's Hospital at Westmead. However, I have been interested in the needs of families with bone and joint challenges since I was a medical student. I grew up in a big family in Melbourne, and while at Monash University I had a great vacation job over a number of years, working as a physiotherapy aide.

My physiotherapist boss worked at a centre for children and adults with long-term disabilities. The assistants needed to help with everything - rolling plaster bandages before casts were made, cutting out elastic support "corsets" for back problems, stitching slings and supports, making little wooden chairs and driving patients to orthopaedic appointments.

We went on home visits, to help carry equipment and to assess people's needs in their own home, school or farm. I learned many facts, but more importantly I learned how people get on with their lives after illness or accidents, and how sometimes simple equipment or social support can make a difference to quality of life. I met families where genetic disorders were present in many members, over several generations, and other families where a new diagnosis had to be faced for the first time.

After medical school, I trained as a paediatrician, enjoying work with many specialists including developmental paediatricians, orthopaedic surgeons and geneticists. I worked in multidisciplinary teams in special schools, in the days before most students were placed in mainstream classes, with extra support.

Committee Profile cont.

I am married and appreciate that having small children of my own taught me a great deal!! We spent time in the USA while they were young, and later I had the chance to retrain formally in Rehabilitation, working in the adult rehabilitation units at Concord and Westmead Hospitals for two years, before returning to the Children's Hospital.

I am able to serve on professional advisory support committees for groups such as ConnecTeD, Osteogenesis Imperfecta Australia, and The Short Statured People of Australia. Our department is very active in training new people in therapy roles, and as paediatric medical specialists. Outside work time, I enjoy family life, gardening, and reading.

ConnecTeD - Friends of The Connective Tissue Dysplasia Clinic - provides advocacy and family support for newly diagnosed patients and their families. The six support groups are:

- Skeletal dysplasias
- Mucopolysaccharidoses (MPS)
- Osteogenesis Imperfecta (Brittle Bone syndromes)
- Marfan syndrome
- Ehlers-Danlos syndrome
- Arthrogyrosis and related disorders

ConnecTeD teamed up with OI Australia to provide the financial support for the Centre for Children's Bone Health and teamed up with MPS Australia and the Jameson Read Trust to form the Centre for Innovative Genetic Therapies.

SPORT'S LUNCH

The third Sports Lunch was held at the Drummoyne Rowers on 21st September 2007.

All the funds raised at these Sports Lunches are for the Clinic.

Special guests on the day were Prof. David Sillence and Dr. Jenny Ault and Karen and Marcus Sykes.

The MC for the day was Warwick Roberts.

Professor Sillence gave the audience an insight into the Connective Tissue Dysplasia Clinic and where the funds raised are allocated. He expressed his gratitude on behalf of the wonderful kids and the Clinic.

Guest speaker, Paul Langmack, provided a sound background into the trials and tribulations of his life and career. Then, surprise guest speaker Willie Mason was interviewed and provided his thought provoking account of the NRL 2007 season.

To top the day off an auction was held for a most diverse range of memorabilia, from a 2005 West Tigers jersey signed by all the team, to a Commemorative item celebrating the Socceroos introduction into the FIFA 2006 World Cup.

Funds raised on the day from ticket sales, raffles and memorabilia auction totalled \$14,182.

This was the third Sports Lunch and funds raised to date including general donations now total \$35,517.

The next Sports Lunch will be Friday 4th April 2008. Guest Speaker, Len Pascoe, former Australian Test cricketer.

If you are interested in attending, tickets are \$85 each or \$800 for a table of 10. Please contact Bill McGill on 0418 965 461 or at billtcb@bigpond.com . Tickets sell quickly.



CONNECTED DONATION SLIP

Yes! I would like to support the Connective Tissue Dysplasia Clinic at The Children's Hospital at Westmead

Please print...

Name (Dr/Mr/Mrs/Miss/Ms): _____

Address: _____

Phone: _____ (home/work) _____ (mobile) Email: _____

Please accept my donation of Please nominate amount \$ _____ \$50 \$100 \$250 \$500

Or instead I would like to make a regular donation of \$ _____ per month per quarter per year

Enclosed is a cheque for \$ _____ made payable to '**ConnecteD – Friends of The Connective Tissue Dysplasia Clinic**'

Or debit my:

Visa Amex Bankcard Mastercard no: _____

Name on Card: _____

Expiry Date: _____

Signature: _____

Please send to: **ConnecteD, Post Office Box 283, Westmead NSW 2145**

Your gift over \$2 is tax deductible