



Connective Issues

September 2009

ConnecteD for Kids

Supporting care for and research into **rare genetic conditions** with fragile bones, skin or blood vessels, stiff joints and short stature

(ABN: 78 484 039 441)

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A New Direction

During the past twelve months, the Committee of Management has come through possible the most intense but exciting time since our inauguration in 2005.

The legal process to become a not-for-profit charity with an independent authority to fundraise, has been a time consuming exercise to comply with all the various legal requirements and requests by the Australian Tax Office, NSW Office of Fair Trading and NSW Office of Liquor, Gaming and Racing. However, all the necessary approval have now been granted.

As you are aware, one of the major changes we needed to make was to expand and broaden our objectives and remove the word "clinic" from our name. Our new name, ConnecteD Connective Tissue Dysplasias Inc, still reflects our main mission to support individuals affected by Connective Tissue Dysplasias (CTD), their families and health care providers.

Trading under the name "ConnecteD for Kids", we will continue to raise funds for the world-class Connective Tissue Dysplasia Service (Clinic) at The Children's Hospital at Westmead. However, at the same time, ConnecteD will be expanding the work it does in others areas, namely to:

- Promote awareness and education about Connective Tissue Dysplasias
- Advocate for people with Connective Tissue Dysplasias
- Liaise with the other organisations that support individual CTD disorders

In conjunction with our 2008 *Linking Together* Ball, ConnecteD launched a Media Awareness Package that has proven to be very beneficial in showcasing the work done by the Clinic. ConnecteD now has its own website (connectedforkids.org.au). The website is still being developed and when it is completed, ConnecteD hopes that it will be a platform for information and discussion on CTD.

Where possible the Committee endeavours to support other CTD organisations and events. Last year members of ConnecteD attended two events on the MPS calendar (AGM and a "morning Tea"). The Non-executive members of ConnecteD (Professor David Sillence and Dr Jenny Ault), supported and attended the conferences of the OI Society, SSPA and International Meeting of the MPS Association. ConnecteD also supported the Muscular Skeletal Meeting for the OI Society of Australia and the Children's Hospital at Westmead.

ConnecteD continues to be indebted to its supporters, and we trust that they will continue that support as we head in this new direction of not only supporting the Clinic but advocating for support and services for all people with Connective Tissue Dysplasias.

REMINDERS:

- **Membership renewal is now due.**

- **AGM**

21st OCT.

Lorimar Dodd's Lecture

Theatre, Level 3, CHW. 7.30pm

Guest Speaker: Dr Craig Munns

- **ConnecteD XMAS STALL**

11th Dec - 8.30 am to 2pm

To be held at CHW.

If you have any goods or can bake cakes etc please contact Lynne Foxall (0405 137 328)

CTD Clinic News

HAPPY



To The
CTD CLINIC

*“Our rehabilitation
and Allied Health
staff have over
800 current
patients”*



This piece of equipment may seem mundane but this extraction pump, purchased from funds raised by ConnecteD for Kids, is vital for the moulding of materials use to make leg and body braces by the Orthotics Department.

Report to the Annual General Meeting from the Connective Tissue Dysplasia Management Service 2008

Next year 2009 will be the 30th Anniversary of the formation of the Multidisciplinary Bone Dysplasia Clinic which became the CTD clinic in 1987 incorporating multidisciplinary services for Marfan syndrome, Arthrogyrosis, Short Stature, Mucopolysaccharidosis, Osteogenesis Imperfecta and Ehlers-Danlos syndrome. Recognising the breath of our service involving both inpatient and outpatients we have renamed the clinic the Connective Tissue Dysplasia Management Service. During 2007 we had over 2500 occasions of service. This last year has been far busier. We continue to be consulted by colleagues in other states particularly Queensland and South Australia and are pleased to have this training role and opportunity to encourage them in service development. When one sees the excellent services provided for all Connective Tissue Disorders it would be unfair to say that we excel in any one are although in the last year we have provided special leadership in Management of Arthrogyrosis, Osteogenesis Imperfecta and Ehlers-Danlos syndromes for which there are no other such services in Australia. Our rehabilitation and Allied Health staff have over 800 current patients.

Arthrogyrosis: This group includes perhaps 200 rare disorders. While the number of patients in current care is small, they need intensive management needs, involving physiotherapy, occupational therapy, rehabilitation and orthopaedic surgery in the first 6-9 months after diagnosis.

Osteogenesis Imperfecta: We have pioneered many areas of care and particularly early management for prevention of back and skull deformity. Dr Munns is pioneering a new method of assessment of bone health known as peripheral Quantitative Computerized Tomography (pQCT). Dr Munns is also assessing treatment of low bone density by the Vibrating Foot Plate enhancement of bone formation. The group has also undertaken molecular genetic diagnosis in patients with OI type VII and type VIII with the assistance of researchers from Baylor College of Medicine in the USA. There is a pressing need for us to set up this service and a research arm in Australia.

Ehlers-Danlos Syndrome: Our hypermobile group has received particular attention from Verity Pacey Senior Physiotherapist and Dr Louise Tofts. Formerly we were unable to properly evaluate “fatigue” and “chronic joint pain” in children with hypermobility. However with Verity and Louise’s efforts we have started to clarify the risk factors for “The Hypermobility Syndrome” and how we might intervene.

Transition of Adolescents to Adult Hospitals: The CTD Management Service has been particularly frustrated by the difficulties in developing services in Adult centres particularly Westmead Hospital despite considerably lobbying to have funding for a centre of expertise recognised in first the Children’s Hospital and then the Westmead Hospital.

Maintaining core knowledge and skills in CTD Management: The Connective Tissue Dysplasia Management Service cares for over 800 current patients with any one of 600 different disorders. The diagnosis is essential to ensure disorder specific care and management. Diagnosis and Assessment is usually carried out by Clinical Geneticists with major input from the Rehabilitation Specialists and our Bone and Mineral Physician, Dr Munns. For many disorders molecular DNA diagnosis is available but from overseas where it is expensive. For example, molecular testing is possible for approximately 200 of the 320 known skeletal disorders. The average cost of molecular genetic testing is over USD 2000. We could achieve so much more for more clients if this testing could be set up in Australia when hopefully there would be considerable cost-saving to Australian families and the health service.

(Continued on page 3)

CTD Clinic News (cont'd)

Education and Training: The clinic personnel with the support of ConnecteD and OI Australia hosted a National Training meeting, *Therapy and its Evaluation in Musculoskeletal Disorders of Childhood* in May 2008. This was an outstanding success with approximately 100 registrants from around NSW and Interstate.

Three members of the clinic attended the 10th International Conference on Osteogenesis Imperfecta. Dr Lanie Alcausin presented a poster on *Growth Hormone Therapy in Children with OI*. Verity Pacey presented a poster on behalf of the clinic on *Multidisciplinary Care for Children with Connective Tissue Dysplasias*. Professor Silence was invited to give the opening presentation on diagnosis of OI.

Future Vision: There are a number of research and development projects whose funding or enhancement would result in significant benefit to our client group and families throughout NSW.

- i. *Therapy intervention in children with Joint Hypermobility* – does a fitness program prevent joint pain and improve mobility. Funding the research program being undertaken by clinic staff particularly the study being undertaken by our Senior Physiotherapist Verity Pacey would help put our physiotherapy research program on a sound footing.
- ii. *Molecular Genetic Diagnosis in Skeletal Dysplasias including OI*. The CHW has a well established molecular genetic service but could take on an expanded role if a senior scientist position was funded (say over 3 years) to develop research methods for rapid DNA sequencing and gene mapping. A research appointment would have the greatest chance of attracting external funding. A major opportunity presented by the proposal of the AGRF to set up a branch molecular genetic laboratory in Westmead during 2009.
- iii. *Training Fellowship or Scholarship in Connective Tissue Dysplasia*. We propose to create a fellowship/scholarship fund to be shared between the departments contributing to the CTD Management Service. There is a very great need for trained personnel in NSW which extends to Australia and New Zealand. This includes the opportunity for therapists to spend periods of time taken as a day a week or blocks of 3 months up skilling in areas of CTD diagnosis, assessment and management. The committee has previously funded in part six fellows, five clinical genetics trainees and one rehabilitation trainee.



In Memoriam

CAMERON

The ConnecteD Committee wishes to express their sympathy to the family of Cameron Sykes, one of the well known and much loved children attending the Connective Tissue Dysplasia Clinic. Cameron was born with Hunter Syndrome (MPS 11), a rare genetic disorder with very complex care needs over many years but sadly a shortened life span. Cameron passed away on the 4th May, aged 10. Our condolences and thoughts go out to Karen, Marcus and Daniel, and to Bill and Dianne McGill and the extended family. Cameron's life was celebrated at a beautiful service which included a reading of the poem *Heaven's Very Special Child*. Cameron was truly one of "Heaven's Very Special" children.

MEGAN

Our deep sympathy also goes out to Caitlyn and Daniel Ly and their family on the sudden loss in June this year, of their three year old daughter, Megan. She was a very bright and active little girl who attended the clinic regularly from babyhood. Megan will be missed by all who knew her.

The Committee would like to gratefully acknowledge the thoughtful and generous support, of both the Sykes and Ly families, in directing donations in memory of Cameron and Megan to ConnecteD for Kids.

Heaven's Very Special Child By Edna Massimilla

A meeting was held quite far from earth,
"It's time again for another birth."
Said the Angels to the Lord above,
"This special child will need much love."

His progress may seem very slow,
Accomplishments he may not show;
And he'll require extra care
From the folks he meets down there.

He may not run or laugh or play,
His thoughts may seem quite far away.
In many ways he won't adapt,
And he'll be known as
handicapped.

So let's be careful where he's sent;
We want his life to be content.
Please, Lord, find the Parents who
Will do a special job for You.

They will not realize it right away
The leading role they're asked to play.
But with this child sent from above
Comes stronger faith and richer love.

And soon they'll know the
privilege given
In caring for this gift from heaven.
Their precious charge, so meek
and mild
Is Heaven's Very Special Child.

Drummoyne Rowers

In August 2006, the Management of the Drummoyne Rowing Club relaunched its "Sportsman's Lunch" fundraisers, and very generously decided that the proceeds of these lunches be donated to support the work of the Connective Tissue Dysplasia Clinic.

The lunches have been very popular and entertaining with a variety of speakers from all sports. Guest speakers have included Steve "Blocker" Roach (ruby league), Doug Walters (cricket), and Malcolm Johnstone (horse racing). In his role as MC, Warwick Roberts has done a wonderful job. Members of ConnecteD have been on hand to lend their support by speaking on the work done by the Clinic and how it is very much appreciated by the families.

The club's restaurant is to be congratulated on wonderful food they provide at these lunches. If you are looking for a lovely meal, overlooking the water, you might consider the *Aqua Vista* (see website for details www.therowers.com.au).

Since 2006, The Rowers have raised over \$70,000 for the work of the clinic. Some of the money has supported extra equipment, some has supported extra therapy hours for patients, and some has gone to updating management guidelines so that children with these rare disorders can have more treatment, closer to their own homes.

ConnecteD would like to take this opportunity to thank all those involved in putting these lunches together. In particular, ConnecteD would like to say Thank-you to Bill McGill, a long time member of the Drummoyne Rowers and a member of our association, for all his help in supporting and raising awareness of the needs of the CTD Clinic.

Support Group News

MPS

(www.mpssociety.org.au)

International MPS Symposium
"Translating research into Clinical Reality"

When: 23 to 27 June 2010

Where: Adelaide

Contact: Wendy@mpssociety.org.au

or see website for further details.

OI Society

(www.oiaustralia.org.au)

OI Christmas Party

Sunday, 29th November 2009, at Auburn Community Picnic Area.

OI 2010 Conference

Friday, 9th to Sunday 11th April 2010, at The Collaroy

SSPA NSW Branch

(www.nsw.sspa.org.au)

Sports Day - October 25, 2009

SSPA NSW will be having a Sports Day at Emerton Leisure Centre, Jersey Road, Emerton (Western Sydney). The day is for all ages, from toddlers to grandparents, boys and girls, short statured or not, regardless of your sporting ability. To help cover costs for the day it will be \$5 per person or \$15 per family.

SSPA Christmas Picnic - November 21, 2009

Date: Saturday 21st November 2009 starting at 11:00am

Venue: Central Gardens, Paton Street, Merrylands.
RSVP: by Thursday 19th November.

NSW Branch Meeting - December 5, 2009

Please see website for further information/contact details for these events.

Fundraising and Activities Report

The ConnecteD Committee of Management would like to thank everyone who has supported ConnecteD during the last twelve months. It has been an exciting time, kicking off with our ConnecteD Linking Together Ball held on 6th September, 2008 and ending with our recent Let's Get ConnecteD Day at Bi-Centennial Park. Both events were very successful in terms of promoting awareness and support for the promotion of Connective Tissue Disorders.

ConnecteD for Kids has been fortunate to have received a number of corporate donations and sponsorships. We would like to thank the following companies for their generosity:

**Access Nursing,
Alcoa Rolled Products,
Bisley & Company Pty Ltd,
Cedarglen Financial Investments Pty Ltd,
Church & Dwight (Australia) Pty Ltd,
Drummoyne's Rower's Club,
Flowers Financial Group,
Franklins Pty Ltd,
Lucy Giumelli Saini Trust,
Regional Area Health,
Smith & Nephew Surgical Pty Ltd,
Wartsila Australia Pty Ltd.**

We would also like to take this opportunity to say thank-you to all the wonderful individual supporters for their contributions, and in particular to Ken and Patty Hatton, who has been our major benefactor for many years. All your donations are important in enabling ConnecteD for Kids to continue its work in supporting those with Connective Tissue Dysplasias.



ConnecteD Linking Together Ball 2008



ConnecteD would like to extend our sincere appreciation to Franklins, for their support. Their generosity has laid the foundations in many directions for Connected for Kids, enabling us to pursue our aims and objectives.

ConnecTeD

Connective Tissue Dysplasias Inc



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Next Issue:

- 2009 Annual General Meeting Reports
- Supporter's profile: Alcoa Rolled Products
- Report on the *Let's Get ConnecTeD Day* held at Bi-Centennial park, Homebush

Annual General Meeting 2009

Wednesday, 21st October 2009

To be held at

Lorimar Dodd's Lecture Theatre, Level 3
The Children's Hospital at Westmead
(take Glass Elevator, main foyer on level 1)

Starting 7.30pm

SPECIAL GUEST SPEAKER

Dr. Craig Munns

Dr Munns, Bone and Mineral Endocrinologist, is a staff specialist at the Children's Hospital at Westmead. Dr Munns is currently involved in both Vibration Research Study and the research into the Deficiencies of Vitamin D in children and adults. Vitamin D deficiency is a major health issue and can contribute to limb pain, fractures, rickets and hypocalcaemic (calcium deficiency) seizures. These issues are very relevant to CTD research.

ALL MEMBERS AND NON-MEMBERS ARE WELCOME TO ATTEND

RSVP by 20th October

Mrs Jennifer Reynolds ph: 9845 6444 or mob: 0409 126 651
or email LynneF@chw.edu.au

THE COMMITTEE OF MANAGEMENT

Like most organisations, the Committee of Management is responsible for the smooth operation of its organisation. The ConnecTeD Connective Tissue Dysplasias Inc is managed by a voluntary committee of nine plus two non-executive professional members. Positions on the Committee are elected on an annual basis at the AGM. Activities of the committee include monthly meeting, overseeing fundraising, promotional and educational events. A small sub-committee is formed for each major fundraising event and activities; sub-committee can include non-committee members.

If you would like more information on the role of the Committee or how you can support ConnecTeD for Kids please contact Lynne Foxall on (02) 9845 3179 or 040 5137 328.