



The OSTEOGENESIS IMPERFECTA Society of Australia Newsletter

June 2007

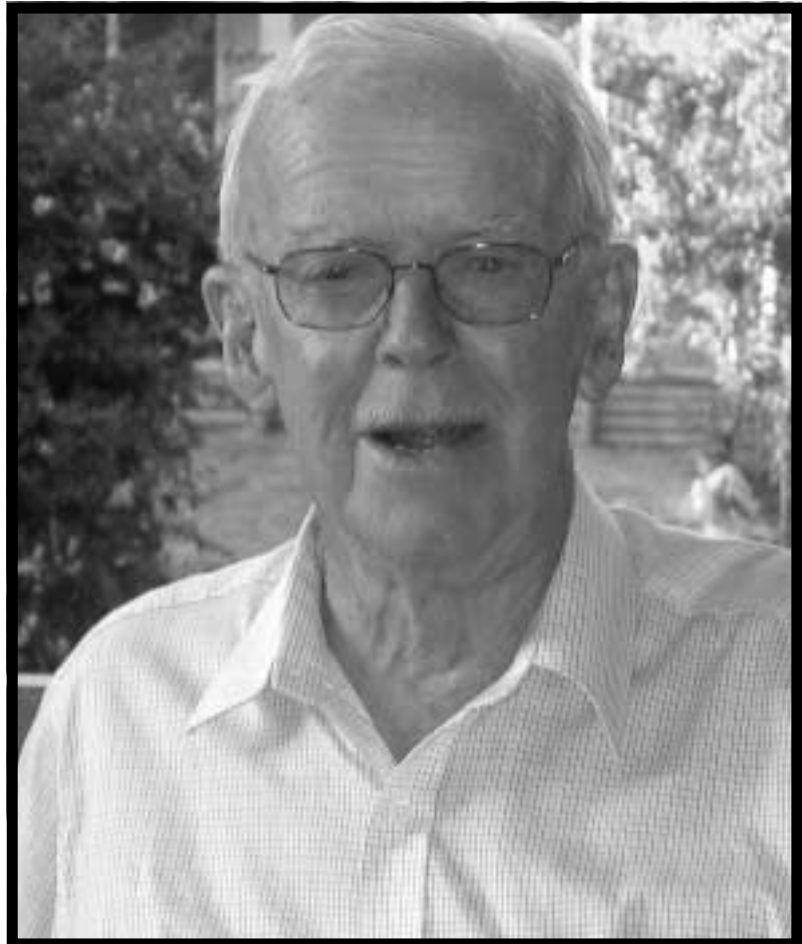
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We're Online!

www.oiaustralia.org



Ian Stuart Foxall Aged 86 years Passed peacefully away at his home 4th December 2006. Ian's contribution, as a Director and member of the OI Society is immeasurable, notably his editorial and fundraising skills. During the last 25 years, Ian has led his family, friends and contacts in supporting the Society, in all its endeavors. He will be truly missed.

Hello Members and Friends,

Our representatives are often the first port of call when a newborn is diagnosed. Our representatives who live with OI become a living example that ordinary life is achievable and are indeed amongst our most valuable resources in the Society! Over on page 5, you'll see an advertisement for a **State Representative (Victoria)**.

We're also seeking a **new secretary** (details also on page 5)

with the departure of our hardworking! Janet Swift (long time secretary) and her husband to Kingscliff, Northern NSW.

On page 7, we are seeking your contributions as to what services, groups etc you've found helpful to you. Input from a range of states or territories are really important to all of us across Australia.

Hope you enjoy this edition.

Craig Andrews
President - OI Society of Australia

Editor Craig Andrews
Email oisociety@oiaustralia.org
Postal PO Box 401
Epping NSW 1710



Research Registry

Dear OI Society,

I am writing from the Kennedy Krieger Institute in the United States because we, with the OIF USA, have established a website for people with OI that lets them record their information and medical histories.

Medical researchers can then use this information by contacting us, and we can provide them with statistics to answer their questions. Or, if they are conducting clinical trials, we notify members in the country the study is taking place in, and provide them with the study and contact information so that each member can contact the researcher if they wish to.

This has already been very successful in promoting research on OI in the United States and Canada, and I am writing to you because we have already have quite a few members who have registered and live in Australia.

Please feel free to look at our site: www.osteogenesisimperfecta.org/oir and if you know of people with OI or researchers that may be interested, we would like to let them know that this resource exists.

Sincerely,
Elizabeth Martin
OI Registry Manager
oirregistrymanager@kennedykrieger.org
www.osteogenesisimperfecta.org/oir

Hi Craig and everybody else,

My name is Bec and I am 9. I have Type 1 OI and I would like to have a penpal, if there is anyone who is around my age. I will be 10 in September.

From Bec
VICTORIA

To reply to Bec - write your letter, put it in an envelope and seal it. Address your letter to **OI Society, PO Box 401, Epping NSW 1710** - and your letter will be sent on to Bec. Don't forget to tell Bec your address! – *Editor*

Travel Help

I am writing to you hoping that you can help me out with some advice. My brother-in-law is coming out to Australia for a holiday from England and he has Brittle Bones.

We are trying to organise hiring a vehicle that would be suitable to fit his electric wheel chair in, such as a people carrier or Toyota Tarago. Would you know any companies that provide this service? Also where could we hire a manual wheel chair?"

MELBOURNE

'Rakem' from Melbourne replies:

After rummaging in my box of tricks, I found a pamphlet for **Wheelaway**
Telephone: 1800 133 320
Website www.wheelaway.com.au
Email wheelaway@personalaccess.com.au

Failing that, perhaps the **Travellers Aid Centre** might be helpful;
Telephone: (03) 9654 2600
Website <http://www.travellersaid.org.au/>
Email info@travellersaid.org.au

Failing that perhaps **Yooralla's Recreation Department** might be useful
Telephone: (03) 9916 5800
Website <http://www.yooralla.com.au/rec.php>
Email: rec@yooralla.com.au

Thank you Rakem for your informative reply. These details will be added to the OI Australia website <http://www.oiaustralia.org/> and kept on file for future queries – *Editor*

Penpals Wanted

Hello,

My name's Kate Cavill and I live with my mum just outside London, England. I'm 22, have OI type 3 and am currently taking some time out from my university degree. I was diagnosed with the condition when I was three days old.

You may know of any members of your society roughly my age, who may be interested in corresponding with me.

Many thanks and best wishes.

Miss K.Cavill
24 Westlyn Close, Rainham, Essex RM13 9JP,
United Kingdom
Kate

1 in 5 Australians

DID YOU KNOW ONE-IN-FIVE AUSTRALIANS reported a disability according to findings from the Australian Bureau of Statistics 2003 Survey of Disability, Ageing and Carers released.

That's almost 4 million people!

Other findings for people aged 15-64 years living in private households include:

1. Many people with disabilities were **not** in the labour force, with participation decreasing markedly with greater levels of core activity limitation.
2. People with disabilities experienced a higher unemployment rate than those without a disability.
3. People with a disability who were employed, were more likely to work in a part-time job than those who were employed and did not have a disability.
4. According to the ABS survey people with a disability were less likely to have completed a higher educational qualification than those without a disability.

This is **not** necessarily to turn you into a raging advocate for the rights of someone with a disability (although go right ahead if that's where you want to be!) – but to let you know you're in GOOD COMPANY!

10th OI Conference

28th, 29th and 30th March '08

Collaroy Conference Centre

Homestead Avenue

Collaroy, Sydney

Contact your State Representative today to start raising funds to meet the costs of attending this conference.

disABILITY EXPO 2007
Adelaide, South Australia

Friday, September 28th
Allan Scott Park
10.30-6.30pm

More information:

mobile: (0418) 804 579

website: <http://www.disabilityexpo.info/>

email: disabilityexpo@yahoo.com.au



We welcome new members to this email discussion group from New Zealand and throughout Australia.


A contact, support and information group for people with Osteogenesis Imperfecta, their families and those who care for them.

<http://groups.yahoo.com/group/OISociety/>

Our email discussion group is

- to offer support and information about living life with Osteogenesis Imperfecta (Brittle Bones);
- to facilitate contact with others who understand your experiences;
- to share links to medical professionals who can care for kids and adults with brittle bones;
- to enable you keep in the loop with state-based OI groups and events;
- and more...

To join, please send a short introduction to the list addressed to oisociety@oiaustralia.org

This group is offered by the **OI Society of Australia** and managed by Craig Andrews on behalf of the Board 



Medicare NEWS –Australia-Wide–

From the first of November 2006, Medicare rebates have been available for GPs to provide early intervention, assessment and management of patients who wish to see a psychologist or psychiatrist.

The initiative known as Better Access to Psychiatrists, Psychologists and GPs is accessible through the Medicare Benefits Scheme (MBS).

More information is available at www.health.gov.au or by calling Medicare on 13 20 11.



Low/No cost Internet Services for people with a disability

“**TADAUST Connect**” - Dial-up internet service for \$5.50 per month. No set-up fee, unlimited downloads and flexible contract arrangements. You must be on one of the following to be eligible: Disability Service Pension (DSP); Aged Pension; or Dept of Veterans Affairs Entitlement Card (subject to conditions).

This service is run by **TADACT in Canberra**.
Payments can be made in advance
3 mths (\$16.50) ; 6 mths (\$33) or
12 months (\$66)
See web site for details or
Telephone **1300 735 439**.

TADAust Connect



Reece Bathrooms –Victoria–

Reece – the supplier of bathroom and plumbing products have opened a **Reeceaccess showroom** in Melbourne as a comprehensive range of accessible bathroom and kitchen products.

The showroom has been designed for users with disabilities.

For more information – visit the showroom
495 Upper Heidelberg Road, Heidelberg West
Telephone (03) 9459-9388
Website www.reece.com.au/access/

The Association of Genetic Support of Australasia (AGSA) is running several different events coming up.



Sibling workshops

- > Tamworth from the 3-4 of November.
- > Surry Hills, Sydney in 2-3 June.

The sibling workshops are run for kids who have a brother or a sister with a genetic condition.



Carers Camp

For a parent and a child without a genetic condition in NSW to spend some quality time together at no cost. 21 – 23 September 2007



Calling Dads!

AGSA wants to find out what can be organised for dads.

Support for families is often geared towards the mum's/female carers – with little focus on the dads.



Telegroup Counselling

- > Siblings group – 23 July to the 10 September
- > Mums group – 4 September to the 23 Oct

If you are interested but not quite sure what is involved – give a Laurie a call, details below.

If you want an invitation to be in on any of these events, call Laurie at AGSA Monday to Weds 9-5pm on (02) 9211-1462 or email projects@agsa-geneticsupport.org.au website www.agsa-geneticsupport.org.au

Vacancy Secretary



AFTER SERVING as Secretary to the Board of the OI Society of Australia since October 2003, Janet Swift will be resigning at the upcoming AGM on 22 September, 2007.

Bernard and Janet have sold their home in Sydney for a sea change move to Kingscliff, Northern NSW.

This leaves a vacancy for **Board Secretary**, which we hope can be filled by someone from within the OI Society of Australia community.

The Board of the OI Society of Australia are seeking a volunteer to assume the role of Secretary and join the productive and committed Board of the OI Society of Australia.

The OI Society of Australia Board is an active, productive team working with medical practitioners, children and adults with *Osteogenesis Imperfecta*, their loved ones and families, to promote greater understanding and care of individuals and families affected by *Osteogenesis Imperfecta*; raising funds for research and provide events and opportunities for contact and support between members of our community.

You must be at least 18 years of age or older; live in the state of NSW; join or be a member of the OI Society of Australia (\$10.00 membership fee), be a team player and motivated to make a difference!

Meetings are held monthly at Auburn RSL.

If you are interested in this volunteer position of Secretary of the OI Society of Australia Board, please contact us by:

email oisociety@oiaustralia.org
phone Lynne Foxall at (02) 9869-1486,
phone Janet Swift on (02) 9871-8283 or
phone Craig Andrews (02) 9566-4110

Before September 1, 2007

Vacancy State Representative (Victoria)



The Board of the OI Society of Australia are seeking a volunteer to assume the role of State Representative (Victoria) and join the productive and committed Board of the OI Society of Australia.

You must be at least 18 years of age or older; live in the state of Victoria; join or be a member of the OI Society of Australia (\$10.00 membership fee), be a team player and motivated to make a difference!

If you are interested in this voluntary position of State Representative of Victoria for the OI Society, please contact us for a Job Description:

email oisociety@oiaustralia.org
phone Lynne Foxall at (02) 9869-1486,
phone Janet Swift on (02) 9871-8283 or
phone Craig Andrews (02) 9566-4110

The OI Society of Australia Board is an active, productive team working with medical practitioners, children and adults with *Osteogenesis Imperfecta*, their loved ones and families, to promote greater understanding and care of individuals and families affected by *Osteogenesis Imperfecta*; raising funds for research and provide events and opportunities for contact and support between members of our community.

Before September 1, 2007

6 ***Osteogenesis Imperfecta Gene Identified***

AN IMPORTANT ARTICLE on the genetics of OI was published in the October 19, 2006 issue of *Cell*, a prestigious medical journal. The research by OI Foundation Michael Geisman Research Fellow Roy Morello, Ph.D., identifies a recessive form of OI. This will greatly improve OI diagnosis and family counseling for people with a family history of lethal or severe OI (similar to Types II and III), or Type VII OI who do not have a collagen mutation.

The research studied a protein called CRTAP (Cartilage Associated Protein) that is important for bone formation. Dr. Morello's mouse studies first showed the importance of this protein for bone, leading the way for human genetic studies.

When the researchers found that CRTAP was in the same chromosomal location as Type VII OI, the genetic connection was made. When CRTAP is in low supply it causes the mild bone fragility of Type VII OI. When it is absent it causes more severe symptoms that are similar to Types II and III OI.

Important discoveries often are the result of teamwork. OI Foundation Medical Advisory Council members Francis Glorieux, MD, PhD, Shriners Hospital for Children, Montreal; Peter Byers, MD, University of Washington; and Joan Marini, MD, PhD, National Institutes of Health; contributed to this research.

Dr. Glorieux discovered the Type VII form of OI. Dr. Byers provided severe OI tissue samples. Independent research from Dr. Marini identified mutations in the identical gene in several cases of recessive lethal OI.

This research provides new information about how OI is inherited for families of children with lethal or severe OI similar to Types II, III, and VII OI. Up to now, research indicated that almost all cases of OI were caused by a dominant genetic mutation in the Type I collagen genes, meaning that only one copy of the OI gene would be necessary for a person to have OI. The gene is either inherited directly from a parent who has OI or results from a new genetic change at the time of conception.

This new research found that lethal or severe OI can also be associated with a recessive form of inheritance caused by changes in the CRTAP gene. In this situation, the parents do not have OI, but unlike in the dominant cases, they carry a mutation for it in their genes. For a child to have OI, he or she must inherit the OI-causing CRTAP mutation from both parents.

This study also provides an explanation for families in which the parents do not have OI, but one or more of their children does. Up until now, this occurrence had been explained as "mosaicism." Mosaicism means that one parent carries the dominant OI mutation in a percentage of the cells that give rise to the sperm or eggs. Parents who have had a child with Type II or III OI have a 2-4% risk of having another child with OI because of this mosaicism.

This new research shows that if parents have a mutation in one of their CRTAP genes, they are not "mosaics". Rather, they are carriers for the newly-identified recessive form of OI. If a child with OI has this new recessive form, then his/her parents have a 25% risk with each pregnancy, of having another child with OI. There is also a 50% chance that each sibling will be a carrier of the recessive gene.

A third area addressed by this research involves the proportion of OI cases caused by collagen mutations. While most cases of OI involve a mutation in the important type I collagen proteins, approximately 10-15% of people who are clinically diagnosed with OI do not have a collagen mutation. This research shows that many (but not all) cases of severe or lethal OI which have a positive biochemical test but do not have a collagen mutation are caused by lack of CRTAP.

This research will lead to several changes in how OI is diagnosed and in the counseling families receive. It is also possible that additional research will find differences in response to treatments between the dominant and recessive forms of OI.

From: OIFoundation, (2007). *Breakthrough*, 32(1), 1, 6. Accessed May 16, 2007 online <http://www.oif.org/site/DocServer/BreakthroughWinter07.pdf?docID=5103>

Contacts Support and Networks

Genetic Support Network –Victoria–

An organisation which aims to maintain a thriving network committed to promoting the interests and well-being of people affected by genetic conditions.

Post PO Box 1100
Parkville VIC 3052
Telephone (03) 8341-6315
Email info@gsnv.org.au
Website www.gsnv.org.au

Genetics Health Services –VIC & TAS–

Provides genetic diagnosis and counselling to the people of Victoria and Tasmania.

Office 10th Floor
Royal Children's Hospital
Flemington Road, Parkville VIC 3052
Telephone A geneticist and metabolic physician are on call at all times and can be reached through the RCH switchboard on (03) 9345 5522
Tasmania (0408) 127 363
Website
www.genetichealthvic.net.au

Women with Disabilities –Australia–

The peak organisation for women with all types of disabilities in Australia.

Post PO Box 605
Rosny Park TAS 7018
Telephone (02) 62448288
Email wwda@ozemail.com.au
Website www.wwda.org.au

Association for Children with a Disability –Victoria–

Information, support and advocacy organisation for families of children with a disability in Victoria.

Post 590 Orrong Road
Armadale VIC 3143
Telephone (03) 9500 1232
Email mail@acd.org.au
Website www.acd.org.au

Genetic Support Network –NSW–

A network of people who are affected by genetic conditions, either directly or indirectly.

Post 66 Albion Street
Surry Hills NSW 2010
Telephone (02) 9211 1462
info@agsa-geneticsupport.org.au
www.agsa-geneticsupport.org.au

Genetic Support Council –WA–

Organisation that acts as a peak body for genetic support groups in WA.

Post Unit 9, 37 Hampden Road
Nedlands WA 6009
Telephone (08) 9389 9377
info@geneticsupportcouncil.org.au
www.geneticsupportcouncil.org.au

Centre for Genetics Education –NSW–

Dedicated to providing current and relevant genetics information to individuals and family members affected by genetic conditions.

Post PO Box 317
St Leonards NSW 1590
Telephone (02) 9926 7324
Email contact@genetics.com.au
Website www.genetics.com.au

Siblings Australia –Australia–

An organisation committed to enhancing the wellbeing of the siblings of children with disabilities and chronic illness.

Post Norwich Centre
Level 1/77 King William Road
North Adelaide SA 5006
Telephone (08) 8361 8361
Email info@siblingsaustralia.org.au
Website www.siblingsaustralia.org.au

WheelAbout –Australia–

Australia's #1 wheelchair accessible vehicle rental service catering for the access transport needs of people with disability since 1998.

Telephone 1300 301 903
Website www.wheelabout.com

Have you a helpful contact, organisation, network or service in your area? Please let OI Australia know about it so we can include it for the benefit of others.



Adult Day & AGM

Sat, 22 September 2007
10am

Wesley Lodge
Hawkesbury Road
Westmead NSW

Young and
Older Adults
Welcome!



13 years and up!
please join us!

We need to know numbers for catering



Views expressed in this publication are those of the individual authors and not necessarily those of the *Osteogenesis Imperfecta Society of Australia*. The material supplied is for information purposes only and not to be used for diagnosis/treatment or any other type of advice. The Editor of the *Osteogenesis Imperfecta Society of Australia* Newsletter reserves the right to edit contributed articles for grammar, spelling or space. OIAustralia accepts advertisements and articles for our Newsletter, although this does not provide an endorsement of these services, products or views expressed. Materials in this newsletter may be reproduced provided the author/s and source is acknowledged.

OI Australia Board

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Janet Swift – **SECRETARY**
– Phone (02) 9871-8283

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Julie Haraksin – **VICE-PRESIDENT**

Mike Kendall – **TREASURER**

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Peter Foxall

Andrew Longhurst

Kristy Melville

Gail Peacock

Christine Taig

PO Box 401
Epping NSW 1710



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Christine Martin – **AUST. CAPITAL TERRITORY**
– Phone (02) 6291-9969

Julie Stonestreet – **NEW SOUTH WALES**
– Phone (02) 9586-4143

No one at present – **NORTHERN TERRITORY**

Raechel Richards – **QUEENSLAND**
– Phone (07) 3841-4057

Abby Myer – **SOUTH AUSTRALIA**
– Phone (08) 8367-9783

Sue Cenin – **TASMANIA**
– Phone (03) 6247-3240

No one at present – **VICTORIA**

Hazel Bellman – **WESTERN AUSTRALIA**
– Phone (08) 9296-3008

