

## Spotlight on the 2012 National Conference Keynote Speaker: Gary Patti

### What's Stopping You?

We're excited to announce that the keynote speaker for the 2012 opening session promises to set an exciting, challenging and motivational tone for this year's conference.

Despite suffering more than 90 broken bones since the time he was two weeks old, and facing multiple challenges throughout his life due to osteogenesis imperfecta,

Gary Patti has never allowed his disability and periods of social isolation and prejudice to stand in the way of achieving what he wants in life.

In fact, Patti says that those barriers only served to motivate him to overcome the setbacks and obstacles with a fierce determination: he dreams big, never gives up, and has the tenacity to go beyond what he thought possible.

Today, Gary owns one of the longest running independent health clubs in the country, as well as an athletic training clinic for youth. He is an ordained minister, motivational speaker and lecturer both in the U.S. and abroad, as well as a former nationally-ranked athlete.

Gary attended the University of Illinois and Rutgers University. He's happily married to his wife, Amparo and resides in New Jersey.

Gary's message is always simple: "What's Stopping You?"

We look forward to having him join us!



## Countdown to Conference! Campaign 2012: Awareness, Advocacy, Action!

The 2012 National Conference on OI, *Campaign 2012: Awareness, Advocacy, Action!* is fast-approaching! The National Conference will be held at the Crystal Gateway Marriott in Arlington, VA, on Friday, July 13 through Sunday, July 15.



The conference is a three-day event filled with informational sessions, social activities, and fun for attendees of all ages! Informational sessions will cover topics such as; Rodding Surgery, Self-Advocacy in Medical Situations, Health Lungs for Children, Nutrition, Building Strong Families, How to Exercise Safely, Mental Health and Advocating for your School-Aged Child. For a full listing of sessions, visit [www.oif.org/conference](http://www.oif.org/conference).

In addition to the over thirty informational sessions to choose from, you won't want to miss the social activities! Sign-up to perform in the Talent Show, attend the Adult's Dinner, or the Kid's Game Night—there is something for everyone! If you would like to perform in the Talent Show, please complete and return the Talent Show Application, which can be found on [www.oif.org](http://www.oif.org).

Registration for the 2012 National Conference on OI is filling up quickly! If you have not already registered to attend this exciting event—now is the time! Visit [www.oif.org](http://www.oif.org) to register! For detailed information about the conference, including hotel and travel information, visit [www.oif.org/conference](http://www.oif.org/conference).

# From the OI Foundation

## Guess the 2014 Conference City!

The location of the 2014 National Conference will be announced at the Awards Dinner and Dance on Sunday, July 15. Use the clues below to see if you can 'Guess the 2014 Conference City!' Send your guess to [conference@oif.org](mailto:conference@oif.org).

1. Don't be fooled by its name, this is not a sleepy town!
2. Ann & Andy would feel right at home at this largest museum in the world.
3. Wear your seatbelt! This is one fast city!
4. They don't serve slippery noodles, but you can order some really old drinks.
5. If you live in the US, there is a 50% chance that you could drive there in one day.
6. Stand by the soldiers and sailors and you'll not only be in the center of this city, but also the center of the state!
7. They'll need a little luck this year.
8. Perhaps the original birthplace of the "top ten" list.

## OI Awareness Week Inspires New Volunteers

Volunteers across the country stepped forward to create new awareness and fundraising activities for our 2nd National OI Awareness Week, which ran from May 5-12, 2012. Our impact is growing!



OI Awareness Week is held in conjunction with Wishbone Day on May 6, an international community awareness effort. Awareness efforts like National OI Awareness Week help educate the greater community, leading to additional funding towards research programs and vital information and resources for our caregivers, doctors, nurses, teachers, and the OI community.

During an eight-day period, members of the OI community held Unbreakable Spirit Walks for OI, music benefits, and *Blue Jeans for Better Bones Days*, and they posted OI Awareness Week posters in communities from Pennsylvania and New York to Oregon and California. Volunteers also reported that both the city of Rochester, NY, and the state of South Dakota each proclaimed May 5-12, 2012 'Osteogenesis Imperfecta Awareness Week'!

In addition, the OI Foundation's *12th Annual Fine Wines Strong Bones* in Arlington, VA, also boasted one of its best turnouts ever! In keeping with the Kentucky Derby theme, participants watched the big Kentucky horse race on May 5 and many donned hats to celebrate the big day. The live auction raised over \$5,000 to support conference scholarships!

It is not too late to hold your own event, either later this year or during OI Awareness Week in 2013! For information, please contact Director of Development Stuart Tart at [STart@oif.org](mailto:STart@oif.org).

## OI Telethon in MA Uses Humor to Build Support

What is an OI telethon like? Well, for two years, Eddie Kane has used a mix of Monty Python-style humor and interviews with both doctors and members of the OI community to build awareness and raise support.

The *Eddie Kane Live* show, based out of Natick and Framingham, MA, held its 2nd Annual Telethon for OI on April 20. The telethon was available for viewing both online and on the local TV channels in Massachusetts. Acts included zany sketch comedy skits, magic tricks, and a variety of musical performances, which aired along with video

# From the OI Foundation

stories from members of the OI community, an in-depth interview with OIF Medical Advisor Dr. Matthew Warman of Children's Hospital in Boston, and the OI Foundation's new promotional video.



Eddie started this telethon in 2011 in honor of his adult sister Kimberlee. **In two years, Eddie's team has raised more than \$3,200!**

Even so, Eddie wrote after the telethon that he is not done fundraising. *"I made a pledge to raise 10,000 dollars, and I am planning four events throughout the summer!"*

## OI Foundation Awards the Continuation of Two Research Fellowships

The OI Foundation is pleased to announce the continuation of Michael Geisman Fellowships to Dr. Christina Jacobsen from Children's Hospital Boston and Dr. Shawna Pyott, University of Washington, Seattle. Each Fellowship awards up to \$50,000 to post-doctoral trainees who are currently working on projects with clear relevance to OI or who have projects will enable them to develop expertise in OI research.

Dr. Jacobsen's research titled, "The LRP5 Pathway: A potential new Therapeutic Target for Osteogenesis Imperfecta" seeks to discover whether a mutation in the LRP5 gene will increase either bone strength or bone mass in mice who have OI. Eventually Dr. Jacobsen hopes to use these mice to find new targets where medication will affect bone strength and bone mass.



**Dr. Christina Jacobsen**

Dr. Pyott's research titled, "Molecular Mechanisms in Recessive Osteogenesis Imperfecta" is focusing on three specific proteins – CRTAP, P3H1 and CYPB – that together form a complex that interacts with collagen. The goal will be to determine how a deficiency of each protein impacts the quality of collagen and thus bone strength and fragility.



**Dr. Shawna Pyott**

Both Dr. Jacobsen and Dr. Pyott are seeing progress in their research and will submit a final document detailing their two year research at this time next year. Dr. Jacobsen's Fellowship has been funded by Mallie's Friends Research Legacy.

## Michelle Hofhine, RN, appointed to NIAMS Advisory Council

Four new members have been added to the Advisory Council of the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), an institute within the National Institutes of Health (NIH). The OI Foundation is pleased to report that Michelle Hofhine, RN, is among this year's appointees. Michelle, who is vice president of marketing for Accredited Home Care Services in Woodland Hills, California, is an active volunteer for the OIF and an OIF support group leader. Michelle has two daughters, Jackie and Nicole. Nicole, who has OI, has participated in several of the research studies on OI at the NIH under the direction of Dr. Joan Marini.

Advisory Council members are responsible for providing advice to the institute on broad policy issues and make recommendations on research proposals.



**Michelle Hofhine**

# From the OI Foundation

## You Can Make a Difference! Spring Appeal Will Fund Research, Information Services

Please help the OI Foundation finish its fiscal year in a strong position on June 30 by contributing to our spring appeal!

This letter, which will arrive in your mailbox in May, offers you the opportunity to direct your support either to research or the Foundation's education and support services.

**Now is your chance to make a difference!** Your contribution to OI research will ensure the OI Foundation has the resources to fund future applications for Michael Geisman Research Fellowships and seed grants for basic or clinical research. A Michael Geisman Research Fellowship provides cash awards to new researchers with doctoral degrees who are currently working on projects with clear relevance to OI, or who have projects that will enable them to develop expertise in OI research. Seed grants are for new or established Principal Investigators who seek to initiate basic or clinical research studies with clear relevance to OI.

In addition, the OI Foundation is partnering on the ongoing OI Adult Natural History Initiative and continuing our support of both the Linked Clinical Research Centers and the OI Registry.

Not only will your gift move forward the research projects described above, they also will

- ◆ Provide information and resources to parents, doctors, educators, and adults with OI,
- ◆ Support the National Conference on OI this July and in the future, and
- ◆ Empower the OI Foundation to continue its advocacy for increased funding from the National Institutes of Health, foundations and corporations.

**Thanks to a Matching Gift Challenge made again by a generous family in Florida, the impact of your donation will be doubled, if you contribute between now and June 30.** You choose whether to support research or the OI Foundation's information and support services for individuals with OI and their families, by indicating your preference on your personalized reply card.

In the OI community, one person or one family can have a tremendous impact. In fact, one volunteer said all he had to do was ask.

Eight years ago, OIF Board member Greg Holman was a father looking for answers that would help his little girl Mallie, who had been diagnosed with OI. He began with a goal to increase the funds available to OI research. By 2011, he and his family had raised more than \$110,000—that was enough money to support a Michael Geisman Research Fellowship for two years!

*"It's up to everyone affected by OI to do whatever they can. ... In the end, we are helping ourselves!"* Greg said.

Please ensure that these important initiatives continue to move forward by making your most generous gift possible today. Thank you for your involvement and support!



**Mallie Holman turned 10 this year. Her diagnosis of OI inspired a campaign by her family that raised enough money to support a Michael Geisman Research Fellowship for two years!**

## Scientists Meet to Discuss OI Related Research and Current Knowledge

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A multidisciplinary panel of researchers and clinicians with expertise in osteogenesis imperfecta met in Chicago, Illinois, April 17-20 for the 12th OI Foundation Scientific Meeting. The meeting, chaired by Dr. Laura Tosi, Children's National Medical Center in Washington, DC, was titled "Assessing the Impact of Osteogenesis Imperfecta on Non-Skeletal Systems." The purpose of the two day meeting was to review current knowledge regarding the non-skeletal consequences of aging with OI, identify major information gaps, and make recommendations to expand the OI research agenda. More than 70 clinicians and researchers attended the meeting.

Prior to the start of the meeting a smaller "think tank" type meeting was held titled, "Molecular Mechanisms, Diagnostic and Therapeutic Strategies." The meeting, chaired by Dr. David Eyre from the University of Washington in Seattle, gathered a group of leading scientists to talk about new research in mineral-collagen interaction; new insights from mouse models and emerging pathogenic mechanisms. Like the larger meeting, outcomes from this two day discussion will help to expand the OI research agenda.

Documents will be available shortly that summarizes the results of both meetings and will be disseminated broadly within the OI research, clinical and patient communities. The OI Foundation thanks the Buchbinder Family Foundation for their continued generous support of this meeting as well the National Institute of Arthritis and Musculoskeletal and Skin Diseases, the Office of Rare Disease Research, the National Institute on Deafness and Other Communication Disorders and Zimmer, Inc for their support.



**The OI-AHNI committee met at the 2012 OIF Science Meeting. From left to right, back row: Dr. Laura Tosi, Mary Beth Huber, Kyle Mulroy, Barbie Simmonds, Dr. Fergus McKiernan. Front row: Dr. Melanie Rak, Dr. Carole Tucker, Annie Kennelly. Not pictured: Lauren Greco, Angela Mancuso, Dr. Mat Oetgen.**

## What to Know Before Traveling Abroad With a Power Wheelchair

A trip to Paris had always been 11-year-old Catie Cheek's dream. She had so often heard stories told by her grandmother of the time spent in Paris just after WWII, the sites she saw and the very night the City of Lights was re-illuminated to mark the war's end.

Those memories left such a strong and indelible impression on her granddaughter that Catie decided to contact the *Make a Wish Foundation* to see if she could honor her grandmother's memories by retracing some of the very sites her grandmother described so vividly. Her appeal proved compelling, and funding was approved for a week's stay in Paris the fall of 2011. Her parents, Jody and Vance would join her.

Both Catie and her dad have OI and use power wheelchairs. Catie's spinal surgery last January requires her to use a power wheelchair while recovering. Jody understood careful planning was required to have the trip go smoothly. She researched charging power wheelchairs abroad online, and followed up with visits to electronic stores such as Radio Shack and Best Buy to confirm what was needed as far as converters or transformers so both wheelchairs would be fully functional and able to be recharged.

However, what she discovered was that no matter how much you plan, you can never over plan. And what you do not know ahead of time could prove costly.

The family arrived at the beautiful Montparnasse section on the Left Bank in Paris on a September morning at 6:00 a.m. By that evening, Vance's chair was drained, and the transformer they purchased did not work, confirmed by a loud pop in the wall. Jody ended up having to push Vance's wheelchair on several outings, and, unfortunately, he was unable to join Catie and Jody when they visited the Louvre and a fashion show on the Champs-Élysées.

After hours of frustrating phone calls, and missed sightseeing time, a rental chair was secured with the help of a couple they befriended whose wife spoke fluent French. However, it was an expense for which the family did not plan on.

"What we learned is the devil's in the details. We had a wonderful time, and Catie was able to see Paris and do the things she loved hearing about from her grandmother, but it would have been a whole lot less frustrating at times had we had the proper information we needed beforehand to charge the wheelchairs," said Jody.

Before planning a trip abroad when using a power wheelchair, Jody offers the following suggestions:

- ◆ Contact the power wheelchair manufacturer directly to learn the voltage requirements and transformer needs for the particular country you plan to visit.
- ◆ Put together a list of people, businesses, organizations and even friends you've met on social media sites who could help you.
- ◆ Contact the OI Foundation for recommendations of contacts abroad.

The following websites are also excellent references for helping you plan for a trip abroad when bringing a power wheelchair:

<http://www.miusa.org/ncde/tipsheets/powerchairs>

<http://www.voltagevalet.com/>



Jody, Catie and Vance Cheek before the Eiffel Tower – September 2011

## Research Update

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Two important recently published articles about osteogenesis imperfecta are now accessible on our website. The chapter *Osteogenesis Imperfecta* by Roy Morello, PhD, and Paul Esposito in the book *Osteogenesis* offers a comprehensive overview of genetics, treatment and recent research. The other is an article by Jay Shapiro, MD, and Emily Germain-Lee, MD, about transitioning care for the young adult who has OI from the pediatric system into the adult health system. This is a topic of critical importance to teens or young adults who have OI, their parents and their doctors. Both articles are free to access and are available on the Breakthrough page of the OI Foundation website under the "Resources" tab.

### Osteogenesis Imperfecta the Book Chapter

Although this chapter is written primarily for doctors, the information is also useful for parents and adults who have OI. This well written chapter condenses a great deal of information from recent research into just a few pages. Some of the highlights include..

- ◆ The authors draw attention to the fact that OI is more than a fragile bone disorder. They include information about the wide range of features that are associated with OI.
- ◆ There is a clear explanation of the genetics of OI including recessive inheritance.
- ◆ The section on classifications looks at the ongoing effort to develop a classification system that reflects the information about OI that has accumulated since the 1970s when the current system of types was established. They explain the need for a classification system to reflect the relative degrees of severity (mild, moderate, severe), genetic inheritance information and the features that require clinical care.
- ◆ The "Management and Treatment" section is detailed and provides clear discussions on the use of drugs from the bisphosphonate family to treat OI and the issues connected to different surgical questions.

The authors conclude that advancements in our knowledge about the genes involved in causing OI improve our ability to develop new treatments. In their opinion one of the next challenges is evaluating the long-term implications of current medical and surgical treatments with the goal of improving the comfort and ability of people who have OI to function as children and as adults.

Dr. Morello is one of the researchers responsible for the discovery of forms of OI that are inherited in a recessive manner. Dr. Esposito is a member of the OI Foundation's Medical Advisory Council (MAC) and an experienced orthopedic surgeon in Omaha, NE.

### Transition of Care

Transition is the term used in medical literature to describe the process of moving a person who has a chronic/ life-long condition from pediatric care into the adult health care system. For people living with OI and other conditions that begin in childhood and last a life-time, transition is often complex, time consuming and very important. In their article *Osteogenesis Imperfecta: Effecting the Transition from Adolescent to Adult Medical Care*, Dr. Shapiro and Dr. Germain-Lee draw on their years of experience. Both authors who are from the Kennedy Krieger Institute in Baltimore, MD, are experienced in the care of people who have OI. Dr. Shapiro is a member of the OI Foundation's MAC. This thought-provoking article is important for parents to read no matter how young their children are, and for parents of teens to read and discuss with their children.

This article presents a set of guidelines for pediatricians, physicians who care for adults, young people who have OI and their families. They present OI as a complex disorder that during the young adult years involves not only unpredictable instances of broken bones, but also the emergence of other health issues such as hearing loss or heart valve disease. Their guidelines point out the importance of four sets of topics.

*continued on page 8*



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#### **RESEARCH UPDATE**

*continued from page 7*

- ◆ Maintaining health
- ◆ Preserving or improving function
- ◆ Continuity of medical and surgical care
- ◆ Psychosocial support

The authors stress the importance of staying informed about the medical aspects of managing OI, especially the symptoms that emerge during late adolescence. Beginning well before age 18, they encourage addressing the physical activity questions that young people face as they begin to live more independently, go to college, and face the rigors of the working world. Continuity of care becomes more complicated as the child with OI becomes too old to be treated at a pediatric center and as his/her health insurance changes. While pediatric care often focuses on forming teams and being comprehensive, the adult system is much more fragmented. This requires the young adult to learn to be proactive about his/her health care needs, to understand their health insurance, and to know how to keep track of their health records. The importance of connecting with new doctors well ahead of an emergency is stressed in this article.

Identifying the issues is an important first step. How to address these needs will differ depending on the severity of the person's OI and the community in which they live.