



BBDC is part of Rare Diseases Clinical Research Network (RDCRN), a program of Office of Rare Diseases Research at NCATS, NIH. The Brittle Bone Disorders Consortium (BBDC) is a group of physicians, researchers, and educators dedicated to learning more about Osteogenesis Imperfecta (OI). We aim to develop new treatments and to improve the overall care of patients with OI by connecting patients with support groups, expert doctors, and clinical research opportunities.



AWARDS

Be the Change

Editorial Message from Eric Rush, M.D.

One of the things I have quickly found out in my adult OI practice is how little we know about the natural history of this disease. It's frustrating as a clinician, because I want to be able to tell my patients what to expect and want to treat them. However, my frustration must only be a fraction of why my adult patients go through in the management of their disease. As the oft-stated and erroneously attributed phrase goes: "be the change you wish to see". And so we must.

In many ways, this is the legacy of rare disease research, which has been classically focused on the natural history and treatment of infants and children. It makes sense, in some ways; in the bad old days when people didn't survive long with rare disease, to focus on younger people. Children's Hospitals were also more used to working with and conducting research on rare disease, so in contrast to research in common diseases, adults with rare diseases were often ignored.

Fortunately, this is no longer the case. There exists much interest in exploring the natural history of rare disease in adults. This is particularly important for adults with OI in whom we expect an extended and in many cases, a normal life span. It is true that many of the things we will learn about adults today won't benefit them directly, but every person has a child, sibling, cousin, or friend who they want to have a better experience. However, some

of what we learn today will be directly translated into targeted or least better therapies and that should be exciting for anyone with OI, even those who are doing well enough.

We need your help to learn more, so I would encourage anyone who is interested to take part in some aspect of the research that is being conducted by the Brittle Bone Disorder Consortium.

A handwritten signature in black ink that reads "Eric T. Rush".

Eric T. Rush, MD, FAAP, FACMG
Clinical Geneticist
Assistant Professor of Pediatrics, Internal Medicine, and Orthopedic Surgery

The Brittle Bone Disorders Consortium

Clinical Sites

Baylor College of Medicine

Principal Investigator: V. Reid Sutton, M.D.

Children's National Medical Center

Principal Investigator: Laura Tosi, M.D.

Hospital for Special Surgery

Principal Investigator: Cathleen Raggio M.D.

Kennedy Krieger Institute / Hugo W. Moser Research Institute

Principal Investigator: Mahim Jain, M.D., Ph.D.

Shriners Hospital for Children, Chicago / Marquette University

Principal Investigator: Peter Smith, M.D. Co-PI: Gerald Harris, PhD, PE

Shriners Hospital for Children, Montreal

Principal Investigator: Frank Rauch, M.D.

Oregon Health and Science University

Principal Investigator: Eric Orwoll, M.D.

University of California Los Angeles

Principal Investigator: Deborah Krakow, M.D.

University of Nebraska Medical Center with Children's Hospital & Medical Center

Principal Investigator: Eric Rush, M.D.

Patient Advocacy Partner

Osteogenesis Imperfecta Foundation:

Principal Investigator: Tracy Hart, C.E.O.

Cores

University of South Florida

Principal Investigator: Jeff Krischer Ph.D.

University of Washington

Principal Investigator: David Eyre Ph.D.

Congratulations to Eric S. Orwoll, M.D, 2016 BBDC PI at Oregon Health & Science University

ASBMR Frederic C. Bartter Award Recipient

The Frederic C. Bartter Award is given to an ASBMR member in recognition of outstanding clinical investigation in disorders of bone and mineral metabolism.

Eric Orwoll is a Professor of Medicine at Oregon Health & Science University. He has been active in basic, clinical, and epidemiological research in bone biology, with a major focus in age related musculoskeletal change, including the epidemiology, etiology and therapy of osteoporosis and sarcopenia in men. His basic/translational interests include the genetics and proteomics of aging. He has been the principal investigator for many projects supported by the NIH, VA and foundations, and is the author of more than 400 peer reviewed publications, reviews, books, and book chapter.



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Meet the OI Community

by Natalie Cinman

Natalie Cinman is a 33-year old female who was diagnosed with Osteogenesis Imperfecta shortly after birth. She is a great spokesperson for the OI community, and at a very young age, published her own article, "A life not so fragile" in the Lancet on her perspective of living with OI. From Montreal, CAN, Natalie shares with us her experiences and hopes for future research.

Q: How is your life experience different from your friends and relatives who do not have OI?

A: Growing up, I did not often feel that I was different and making friends was never an issue. I have always felt that people perceive you the way you perceive yourself, so I always made an effort to be approachable and friendly. Overall, I feel that I got to experience a lot of things that I would not have, had I not had OI.

Q: How has your perception/outlook on OI changed as you have gotten older?

A: As I get older and am enjoying the life I am currently living, my perception has not really changed, as I have not changed. I am still the outgoing person I have always been. This is one of the traits that my husband said attracted me to him. That I made him feel comfortable.

Q: You have been, and continue to be, a great spokesperson for the OI community. What message would you like to relay to others living with OI?

A: I believe that you need to be true to yourself. No one knows your body and your limits better than you do. Be an advocate for your medical care; teach the world both socially and in the medical profession about OI. People are always leery about things they don't know. Try not to get frustrated when people ask questions because you are educating them.



Natalie Cinman
and husband
Denis Toupin

Q: As a previous participant in the LCRC, now transitioning into the BBDC, what prompted you to participate in research, and why should others participate?

A: I believe that continuing research on OI is imperative for our generation and future generations. Given the medical advancements, the prognosis is no longer so black and white. Being part the first generation of people to receive the pamidronate treatment, it is important to monitor how we age and the long-term effects. As a woman, progression through menopause is an important thing to monitor too. Also, educating medical professionals.

Q: What future or additional research projects do you think would most benefit individuals with OI?

A: Studies about heart function, not just pulmonary function, would be very beneficial. Heart/lung compromise are the most common ailments in people with OI. Also, a study on physical therapy for people given their various limitations. As a woman, I would be interested in a study on what menopause will look like in the future, what will be of our bones, and how it will affect us hormonally. Studies on weight management and its effect on our bones given the sedentary lifestyle of individuals with OI would be interesting also. Recurring pain and pain management comes up a lot on Facebook groups for people with OI. We are all aging and this is something that might be of interest as well.

Advocacy Partner

by Petra Harvey, Health Education and Outreach Manager at OIF

New Resources from the OI Foundation

Materials for Adults with OI and Their Doctors

At the OIF National Conference a new set of materials was introduced:

Take Charge of YOUR Health.

This tool kit is designed to help adults partnering with their doctors to lead healthy lives. The materials include a **Pocket Guide** available from the OIF online store and the following fact sheets that are posted on the OIF website www.oif.org, under the tab "Especially for Adults."

For All Adults

- [Take Charge of Your Health](#)
- [Take Charge of Your Breathing](#)
- [Pneumonia Vaccine Update](#)
- [Poster: Handle with Care. Adults](#)

For Teens and Young Adults

- [Moving from Pediatric to Adult Care](#)
- [Know Your OI: A Basic Health History](#)

To Share with Your Doctors

- [Care for the Adult Patient with OI](#)
- [Chart: OI and Common Medical Tests and Procedures](#)
- [Resource for Medical Professionals](#)

If you would like to receive a hard copy of these materials, please email Bonelink@oif.org or call the office at: (301) 947-0083.

OI REGIONAL CONFERENCE UPDATE

The OI Foundation sends a special thank you to the speakers, attendees and volunteers of the OIF Regional Conference in Houston! The one day meeting, chaired by OI Foundation Medical Advisory Council Member Dr. Reid Sutton, was held at Texas Children's Hospital Pavilion for Women on Saturday, November 12, 2016. This meeting welcomed approximately 100 individuals from the OI community around the region. Families traveled from as far as Atlanta, Georgia to attend this meeting.

Speakers included Dr. Brendan Lee of Baylor College of Medicine, Dr. Reid Sutton of Baylor College of Medicine and Texas Children's Hospital, Dr. Cathleen Raggio of the Hospital for Special Surgery in New York, Dr. Sandesh Nagamani of Baylor College of Medicine, Dr. William Phillips of Texas Children's Hospital, Dr. Vinitha Shenava of Texas Children's Hospital, physical therapists Stacy Bucic and Emily Hermes, and licensed mental health counselor Dr. Michelle Fynan.

This year, the OI Foundation is gearing up for two regional conferences. The next OI Foundation Regional Conference will be held at the Shriners Hospital for Children - Montreal, Quebec, Canada on June 3, 2017. Visit the [OIF Regional Conference page](#) for updates as they become available.



Upcoming Events

- OIF Scientific Conference
April 20-21, 2017 in Chicago, IL
- American Society for Bone Mineral Research
September 8-11, 2017 in Denver, CO

Consortium Activities

FOA for pulmonary study

In late 2015 the OI Foundation began a multiple pronged effort to address the issue of the seeming rise in adults with OI experiencing pulmonary complications after and during "routine" medical procedures; experiencing severe pulmonary complications while battling a respiratory illness; and experiencing an increased dependence on supplemental oxygen to help keep lung function at a productive level. The OI Foundation and a group of dedicated OIF volunteers have raised funds to conduct research into this area.

Questions may be directed to thart@oif.org or telephone 301-947-0083.

Addition of new sites (US and international)

Data access

DID YOU KNOW?!

WHAT IS COLLAGEN?

Collagen is the substance that holds the body together. It is present in many tissues other than bones, like the eyes, blood vessels, and teeth.

WHY BLUE SCLERA?

The sclera appears to be blue-grey in some individuals with OI because of the abnormal collagen present in the eyes. The abnormal collagen causes the sclera to be thinner, allowing the darker colored tissues underneath the sclera to show through.

OTHERS WITH OI?

The actor, Atticus Shaffer, seen in the hit TV program *The Middle*, has Osteogenesis Imperfecta

Meet the Doctors

Frank Rauch, M.D.

OI is a rare condition, but it certainly does not feel that way when you work here at the Shriners Hospital for Children in Montreal. The OI program in our institution was established 25 years ago through a collaboration between Dr. Francis Glorieux in genetics and Dr. François Fassier in orthopedics. Ever since, OI has been a big topic in this hospital, both in clinical care and in research. In the 15 years that I have been working in Montreal as a pediatrician and as a scientist, I have seen more than 650 children, adolescents and young adults with OI (and I have learned to say 'OI' in many different languages!). Thanks to the generous support from the Shriners organization, many of my patients have been able to participate in studies on a variety of OI-related topics, such as finding new genetic causes of OI, the role of vitamin D supplementation, and evaluating the long-term effects of bisphosphonate treatment, to name a few recent study topics.

Working with all these youngsters with OI has been an amazing journey. People that I first met when they were toddlers are now about to enter university. Many that I treated as teenagers now have children on their own. Realizing this makes me feel old, but more philosophically, watching patients grow up and listening to what they say makes doctors learn a lot. That knowledge can be used to help the next generation of patients. What is true for the individual doctor is also true for medical science as a whole. This is what studies like the Brittle Bone Consortium are about. We are collecting information on how people with OI are doing, and this will bring new insights that eventually feed back into improved treatments.

And we certainly need improved treatments. True, bisphosphonate treatment helps to make the bones stronger and decreases the number of fractures. Tweaking the treatment protocols has simplified things. When I first prescribed bisphosphonate treatment, kids each year spent 9 full days in hospital to receive the infusions. Now, they need to come in only for one morning every 6 months. There also have been many improvements in surgical procedures since I

started working on OI. But these therapies leave a lot of room for improvement. Many kids with OI still have a lot of fractures and their growth is often slow. We do not know much about how to treat adults with OI. And for many aspects of OI, such as dental issues and lung problems, we are just starting to think about treatments. Working together in the Brittle Bone Consortium gets people on the same page and helps to find out what works best.



Frank Rauch, M.D.
Professor of Pediatrics, McGill University
Shriners Hospital for Children

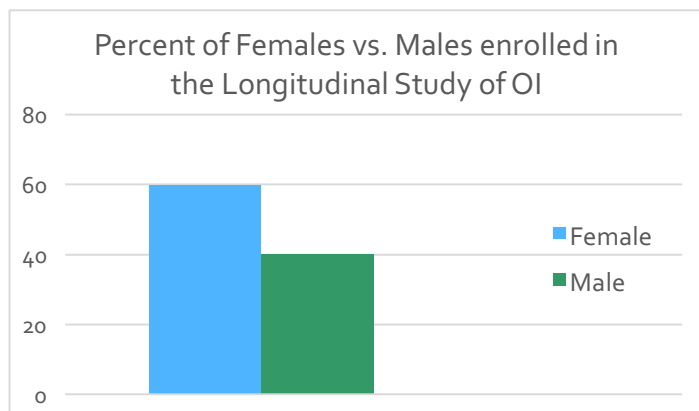
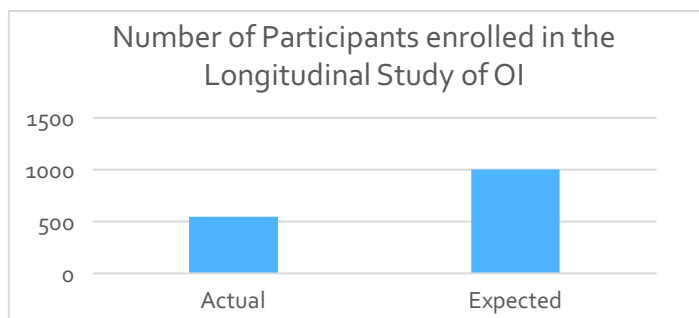


CLINICAL RESEARCH STUDIES

CURRENT:

Natural History Study (BBD7701) With the overall aim of this study being to enhance the quality of life of those with OI, we are currently performing a long-term follow-up of a large group of people with OI.

[Check out statistics on the Study Below!](#)



Biomarker Study

The goal of this study is to develop a non-invasive test that can determine OI-subtype. You must be enrolled in the natural history study to be eligible for this study.

[Statistics on the biomarker study:](#) 12 participants have been enrolled!

Dental Craniofacial Features of OI (BBD7704)

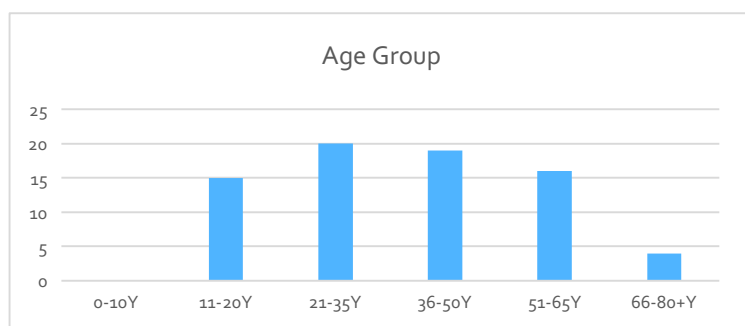
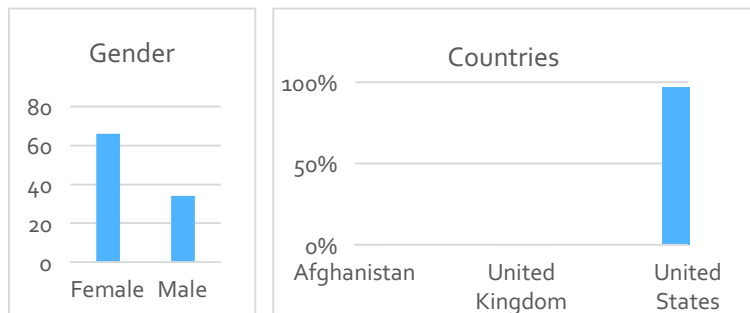
The purpose of this study is to find out more about the facial and dental structures of individuals with OI. You must be enrolled in the Longitudinal study to be eligible for this study. We will perform a cone-beam-CT of your jaw and correlate the physical findings with your genetic findings.

For more information on the above studies, please visit:
<https://www.rarediseasesnetwork.org/cms/bbd/Get-Involved/Studies>

Contact Registry

The Rare Diseases Clinical Research Network (RDCRN) Brittle Bone Disorders Consortium (BBD) Contact Registry is a way for patients with brittle bone disorders and their family members to learn about BBD research studies they may be able to join. Contribute to knowledge about rare diseases by answering questions about yourself, your health, and your quality of life.

Statistics from the Contact Registry



For more information, please visit:

<https://www.rarediseasesnetwork.org/cms/bbd/Get-Involved/ContactRegistry>

PROMIS study tool (BBD7702)

The PROMIS tool is a self-reporting on-line survey allowing individuals with OI to report their quality of life. We plan to compare the self-reporting results with the clinical data collected in the Longitudinal study to validate the PROMIS tool. This study is currently underway.

UPCOMING:

Pregnancy Survey (BBD7705)

The purpose of this study is to learn more about the course of pregnancy and birthing outcomes in women with OI. All women with OI are eligible to take this online survey. This study will be ready for enrolment early 2017.

Drug Therapy Trial (BBD7706)

This clinical trial will help us determine if a new drug is helpful in the treatment of OI. The first phase of this trial will focus on drug safety and tolerability. We will be recruiting and enrolling adults with severe OI Spring 2017.

How to Participate:

Use the following links:

[Participating Clinical Centers](#)

[Get Involved](#)

[ClinicalTrials.gov](#)

[Osteogenesis Imperfecta Foundation](#)