

The OI Foundation's Patient-Centered Outcomes Research Project Continues

The OI Foundation is well underway with our project, **Preparing Adults with Osteogenesis Imperfecta to Engage in Research on Access and Quality of Care for Their Rare Disease**, which has received an award from the Patient-Centered Outcomes Research Institute (PCORI). This project's many ventures include not only the listening sessions announced in previous Breakthrough issues, but also interviews with leaders of both adult and pediatric OI clinics, leaders of professional medical and nursing organizations, healthcare transition researchers, and leaders of patient advocacy organizations. Interviews currently in progress include leaders of

the Rare Bone Disease Alliance, who have graciously volunteered to contribute their expertise.

Topics explored in both the listening sessions and interviews include the healthcare needs of young adults with OI transitioning from pediatric to adult care, as well as identification of barriers and opportunities for accessing quality healthcare across the age span. We particularly seek to identify innovative



Patient Advisory Board Member Michelle Fynan, PhD with her family

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“Many individuals with OI struggle to find medical professionals with current knowledge of the concerns presented by OI. For someone aging out of pediatric care this may coincide with other significant life changes such as higher education, entering the workforce and possibly independent living outside their family. . .”

**OIF Regional Conference:
Washington, DC**
JUNE 10, 2023



Save the Date!

On Saturday, June 10, 2023, the OIF will host a **Regional Conference in Washington, DC!** This one-day meeting will feature presentations by local speakers and OIF Medical Advisory Council (MAC) members. OIF Regional Conferences are opportunities for local community members to meet each other and hear from expert physicians. The meeting will take place at the Westin Crystal City at Reagan National Airport. More information and a link to registration can be found on the OIF's events page at www.oif.org/events.

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strategies that the OI Foundation might employ to help meet the needs of our growing adult community. We have continuously engaged the project's Patient and Scientific Advisory Boards to help advise and give us feedback during the creation of the guides and scripts we use for these discussions.

"Many individuals with OI struggle to find medical professionals with current knowledge of the concerns presented by OI. For someone aging out of pediatric care this may coincide with other significant life changes such as higher education, entering the workforce and possibly independent living outside their family. The PAB has recently focused on brainstorming recommendations and guidance for the OIF to assist with the transition from childhood to adult care with a focus on these other changes. I believe the experiences and ideas being gathered by those affected by OI are important and will contribute to better tools in the future," says Barbie Simmonds, a member of our Patient Advisory Board (PAB).

Another PAB member, Michelle Fynan, PhD, states "In my experience growing up with OI, my childhood and adolescence was heavily focused on fracture care, corrective surgeries, and rehabilitation. Then, when I stopped growing and the fractures

were fewer, there was this illusion of not needing to focus on my health into early adulthood. But now that I am aging with OI and seeking out quality care, I'm currently facing challenges in finding physicians in my area who will work with me. I'm hopeful that this project will help prepare future generations of individuals with OI, including my daughters, so they can navigate this transition into adulthood with a solid plan for continuity of care."

We are still looking for individuals to participate in the listening sessions who are 18+ living with OI, as well as parents and caretakers, from the Northeast, West, and Midwest United States.

During your listening session you will brainstorm with 5-7 other members of the OI community for about 90 minutes on the topics outlined above. Each listening session will be conducted virtually via Zoom and sessions will continue to run through the end of this year. Consistent with PCORI policy, we will compensate all participants for their time.

Please contact us if you are interested in participating in one of our listening sessions or if you have any questions by reaching out to Stacie Connors at sconnors@oif.org.

Golf Events for the OI Foundation

Thank you to the many wonderful families who hosted golf fundraisers in support of the OI Foundation this year. Teresa & Ken Gudek and OI Foundation board member Jim Early and his wife Jane hosted the Matthew Smith Memorial Golf Outing at Manchester Country Club in Manchester, NH; the Walls family hosted the Unbreakable Spirit® Golf Outing at the Iron Valley Golf Club in Lebanon, PA; and the Abruzzi family hosted the Abruzzi Golf Tournament at the Willow Hollow Golf Club in Leesport, PA. We are so grateful for your support!

For details about all upcoming events, you can visit www.oif.org/events.



Jim Early (l) and Ken Gudek (r) were joined by members of OIF Founder, Gemma Geisman's family at the Matthew Smith Memorial Golf Outing

A Big and Bright Night in Houston, TX

The 6th annual Boots & Bling for Better Bones was held on Saturday, October 15th in Houston, TX. Host committee member Cindy Medina was joined by her son, Matthew, to tell their OI story to the nearly 150 attendees who came together to raise \$105,000 for the OI Foundation. Attendees enjoyed the legendary Goode Company barbeque and live music while bidding on fantastic auction items. To view photos from this event, please visit the OI Foundation Facebook page.





Season's Greetings

Thank you for thinking of the OI Foundation this holiday season!



Thank You for Thinking of the OI Foundation This Holiday Season!

Find the perfect gift for everyone on your list while supporting the OI Foundation!

1. Shop online using AmazonSmile for the OI Foundation! For every online purchase you make, Amazon will make a donation to the OI Foundation. Shop at <http://smile.amazon.com/ch/23-7076021>.
2. Gift *Unbreakable Spirit*® apparel to friends, family, and yourself! Shop the OIF's Online Apparel shop at www.oif.org/shop.
3. Make a donation in honor or in memory of a loved one to the OI Foundation at www.oif.org/donate.

For more information about AmazonSmile, the OIF online store, or stock giving, please contact the OI Foundation at bonelink@oif.org.

The Current State of OI Research

The OI Foundation's *Current State of OI Research* video series was created to highlight and describe the work of the Brittle Bone Disorders Consortium (BBDC). The OIF has hosted six sessions in this series so far. During the most recent session, the OI Foundation discussed the Longitudinal Study (also known as the Natural History Study) with Dr. Reid Sutton (Geneticist at Baylor College of Medicine, Administrative Director and Principal Investigator of the Brittle Bone Disorders Consortium (BBDC), member of the OIF Medical Advisory Council and Board of Directors). Dr. Sutton discussed the design of the study, its goals, and what researchers have learned so far.

To view session recordings, please visit the OIF website or the OI Foundation's YouTube channel at www.youtube.com/@OIFoundation.

Participate in Current OI Studies

Often, the success of clinical studies of a rare disorder like osteogenesis imperfecta (OI) depends on getting enough people to participate in the study so the results are meaningful. If you are interested in learning more about current OI studies or would like to join the OI Registry, please visit www.oif.org/currentstudies.

GET IN TOUCH WITH THE OIF



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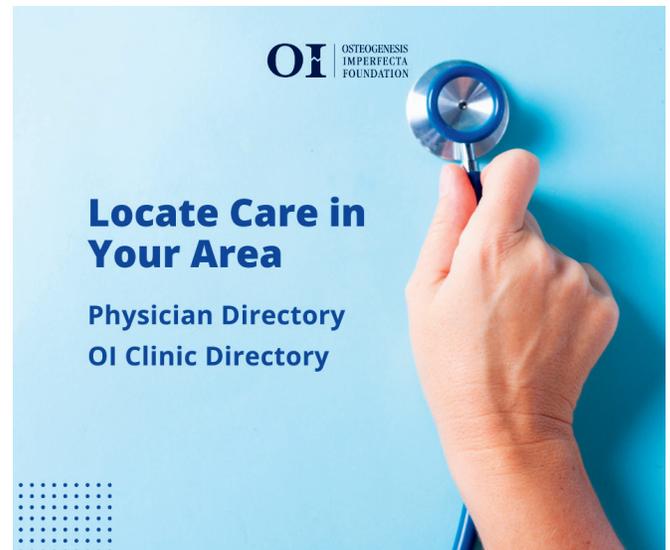
Edward D. Cranston College Scholarship

The OI Foundation is excited to announce the creation of the Edward D. Cranston College Scholarship. This scholarship is in honor of Edward D. Cranston, who was born in 1962 with severe osteogenesis imperfecta. Edward challenged himself by pursuing all educational opportunities available to him to achieve his goal of working as a health care professional. He dedicated his career to working with children with complex respiratory conditions and is held in high regard both by professionals in the field, as well as the families of the many children with whom he has worked. He continues volunteering with school and community groups, raising awareness about disability.

This scholarship is for students with osteogenesis imperfecta who are applying to or have been admitted to a college or university. Applicants must articulate a need for scholarship assistance to meet their academic goals, be committed to full-time college enrollment and show proof of enrollment in college to receive the funds. The scholarship is intended for beginning college freshmen. Preference for the scholarship is based on a combination of merit and financial need. Applicants are required to submit at least two letters of recommendation from current high school teachers. Applications will open in the new year, so stay tuned for more information.

Jeanie Coleman Impact Grant Program

The Jeanie Coleman Impact Grant program provides direct support to individuals and families living with OI in need of equipment or services that will improve their quality of life. Items that have been awarded in the past include (but are not limited to) wheelchairs, technological devices (laptops, tablets), hearing aids, home adaptation, and accessible vans. Applications for the 2023 cycle open on January 9th, so stay tuned for more information. If you have any questions about the program, please feel free to reach out to Stacie Connors at sconnors@oif.org.



Locate Medical Care in Your Area

The OIF Information Center maintains a list of medical professionals (ie. general physician, dentist, specialist) who have experience treating patients with osteogenesis imperfecta. To locate care in your area, please email bonelink@oif.org.

The OI Foundation also maintains a list of hospitals and clinics that offer coordinated treatment for people living with OI and other bone disorders. Visit the OIF's Clinic Directory at www.oif.org/ClinicDirectory.





As we near the end of 2022, we are taking a moment to reflect on the accomplishments of the OI Foundation this year. We are thankful for the generosity of individuals who have allowed the OI Foundation to continue providing programs and services that benefit all members of the OI community. Please take a moment to help the OI Foundation continue our efforts to improve the quality of life for individuals with OI by making an end of year contribution using the enclosed envelope or visiting www.oif.org/donate. Thank you in advance for your support!

Eugene Washington PCORI Engagement Award

Under the direction of OIF Medical Advisory Council member Dr. Laura Tosi, the OI Foundation recently completed our 3rd Eugene Washington PCORI Engagement Awards, through the Patient-Centered Outcomes Research Institute (PCORI), *Leveraging Virtual Communication to Advance PCOR Adoption by the Rare Bone Disease Community*. This award brought together members of the Rare Bone Disease Alliance (RBDA) with stakeholders from the patient and scientific communities in a series of focus groups and large group meetings to examine barriers to participation in research, barriers to access to care, and patient-identified issues that need to be reflected in the development of patient-reported outcomes. The OI Foundation is currently administering our latest award, *Preparing Adults with Osteogenesis Imperfecta to Engage in Research on Access and Quality of Care for Their Rare Disease*, which is exploring the problems that individuals with OI have as they transition from pediatric to adult care. Over the next year, we will be hosting more than 150 listening sessions with members of the OI Community, Clinicians, and other rare disease stakeholders to explore challenges a patient may encounter when seeking primary and specialty care, the difficulties pediatric OI clinics have as they seek to successfully transition their patients to adult care, and the barriers that existing adult OI clinics encounter as they seek to provide appropriate and high-quality care to adult patients.

Scientific Meeting

In April, the OI Foundation gathered 70 scientists and researchers specializing in OI research and treatment in person – with an additional 65 attending virtually – for a two-day session to discuss *Scientific Advances in OI*. This meeting is designed for scientists interested in learning more about the latest in basic and clinical OI research, and how the information could ultimately benefit people living with OI. This year's meeting, chaired by Dr. Deborah Krakow, featured sessions on the musculoskeletal system, quality of life, stem cell research, cardiopulmonary issues, treatment beyond bisphosphonates and a look at what's new on the horizon.

Virtual Education and Mutual Support Sessions

For the past three years, the OI Foundation has hosted more than 20 live, virtual sessions connecting OI community members with OI experts to discuss COVID-19 related topics, research updates, and mutual support opportunities. Sessions have included: three Virtual National Conferences; Information on COVID-19; Mental Health and Wellness; Nutrition and Fitness; Employment Accommodations; COVID-19 Vaccine Updates; Brittle Bone Disorders Consortium research updates; support groups, virtual holiday parties and so much more! The virtual sessions have allowed hundreds of new individuals who have not had the ability to attend in-person OIF events to participate in the most popular informational and social sessions. All OIF virtual sessions have been recorded and are available on the OI Foundation's website and YouTube channel with certain key educational sessions being available in English, Spanish, and French.

Rare Bone Disease Alliance

The Rare Bone Disease Alliance (RBDA), a program of the OI Foundation, brings together a coalition of sixteen rare bone disease patient advocacy groups. The RBDA is committed to expanding education of rare and ultra-rare disorders and expanding research on the natural history, diagnosis, and treatment of the diseases. The RBDA

unites researchers and early-stage investigators through a monthly TeleECHO Clinic Series that focuses on helping physicians, nurse practitioners, physician assistants and other healthcare professionals safely and effectively diagnose and treat rare bone diseases and disorders. The RBDA also hosts scientific symposia and working group meetings through associations such as the American Society for Bone and Mineral Research and the American Society of Human Genetics. In 2023, the RBDA will host a scientific symposium led by Dr. Eric Orwoll focused solely on osteogenesis imperfecta.

OI Clinic Outreach

In an ongoing effort to connect with medical professionals who are currently treating patients with OI and educate them on the resources the OIF provides clinics and their patients, the OIF has begun holding a biannual Town Hall Meeting for OI Clinic and Bone Health Programs with the first two events being held in April 2022 and November 2022. We were excited to gather medical professionals in person and virtually to connect and collaborate on the latest in OI treatments and patient care. We have created a new quarterly electronic newsletter specifically for this group, which includes important announcements about meetings and resources.

OIF National Information Center

This past year, OIF staff responded to more than 11,000 direct inquiries for medically verified information, and an average of 6,000 people used the OIF website each month. Topics range from medical issues such as genetics, diagnosis, and treatments to daily living strategies such as school and employment. Educational materials are available in print and electronically through the OIF website.

Social Networking

The OI Foundation manages three official social networking sites: the OIF Facebook Page – followed by 14,257 Facebook users; the OI Foundation Twitter page (@OIFoundation) – followed by 2,786 Twitter users, and the OIF Instagram page (@oifoundation) – followed by 1,804 Instagram users. This year the OI Foundation OIF YouTube channel highlighted virtual information sessions, COVID-19 community calls, and fundraising event recordings. A new playlist was created in honor of OI Foundation founder, Gemma Geisman, to share stories from OI community members. *Gemma's Corner* encourages the OI community to keep sharing with each other, caring for each other, and providing support to one another.

Jeanie Coleman Impact Grants

In partnership with the Children's Brittle Bone Foundation, the Jeanie Coleman Impact Grant Program was designed and established to provide funding for items that will significantly improve the quality of life for a person who has OI and limited financial resources. In 2022, the OIF was able to fund \$136,509 worth of items and services for 18 families. The 2022 grants covered a range of items including: mobility assistive devices, car seats and ASL class.

The next round of Impact Grant applications will open in January 2023.

With your help, the OI Foundation has accomplished so much over the years, but there is so much left to do. We cannot continue providing the programing and information members of the OI Community have come to rely on without your support!

OI Foundation Attends Medical Professional Meetings

The OI Foundation is grateful to be able to attend medical professional meetings to connect, educate, and learn from researchers, scientists, and physicians in the rare bone disease field from around the world.

This fall, representatives from the OI Foundation attended the American Society for Bone and Mineral Research (ASBMR) 2022 Annual Meeting in Austin, Texas. The weekend featured more than 100 informative sessions, 1,000+ poster presentations, and many opportunities to connect with new and old colleagues. During the meeting, OIF CEO Tracy Hart co-chaired the 'Challenges in Clinical Trials' session alongside Dr. Frank Rauch (OIF Medical Advisory Council Member) during the 2022 Symposium. Also, OIF Medical Advisory Council member Dr. Brendan Lee was awarded the William F. Neuman Award. We are honored that he recognized his patients, OIF CEO Tracy Hart, and the OI Foundation during his acceptance. Congratulations, Dr. Lee!

The OI Foundation and Rare Bone Disease Alliance also attended the American Society of Human Genetics Annual Meeting in Los Angeles, California. Our group hosted an ancillary meeting titled *Treatment of Rare Bone Diseases from Bench to Bedside*, co-chaired by Dr. Carlos Ferreira (National Institutes of Health (NIH)) and Dr. Sandesh Nagamani (Baylor College of Medicine). This meeting was attended by 50 medical professionals. The agenda featured presentations on Fibrous Dysplasia, Achondroplasia, Hypophosphatasia in Adults, and X-linked Hypophosphatemia.

We look forward to attending and hosting future medical professional meetings to help move OI research and education forward.



OIF staff members with William F. Neuman Awardee Dr. Brendan Lee

Upcoming Events for Medical Professionals

On Friday, June 9, 2023, the OIF is hosting an **OI and Bone Health Clinic Town Hall** in Washington, DC at the Westin Crystal City at Reagan National Airport. These biannual meetings provide the opportunity for medical professionals treating patients with OI and those who are interested in bone health to connect and collaborate, as well as learn about OIF resources. More information on this event can be found at the OIF's Medical Professional Calendar at www.oif.org/medprocalendar.

The **Rare Bone Disease TeleECHO Clinic Series** continues to hold sessions in its fourth year. This monthly virtual seminar is for medical professionals who want to learn more about diagnosing and treating rare bone diseases beyond just OI. Each session includes didactic presentations and group discussions. To view the full agenda and register to attend upcoming sessions, visit www.rbdalliance.org/echo.



Rare Bone Disease

Thursday, January 5, 2023; 3:00pm EST

Clinical Spectrum of ENPPI and ABCC6 Deficiency

David Weber, MD, MSCE, *Medical Director, Center for Bone Health, Children's Hospital of Philadelphia*

Thursday, February 2, 2023; 3:00pm EST

Calcium Sensing Disorders

Rajesh Thakker, FRS FMedSci FRCPATH FRCPE FRCP, *May Professor of Medicine in the Nuffield Department of Clinical Medicine at the University of Oxford and a fellow of Somerville College, Oxford University of Oxford, Radcliffe Department of Medicine*

Thursday, March 2, 2023; 3:00pm EST

Gene Therapy for Repair and Regeneration of Bone and Cartilage

Brendan Lee, MD, PhD, *Professor and Chairman of the Department of Molecular and Human Genetics, Baylor College of Medicine*

Thursday, April 6, 2023; 3:00pm EST

Non-OI Disorders of Collagen

Geert Mortier, *Director of the Department Medical Genetics and Head of the Center for Rare Diseases in the Antwerp University Hospital, Chairman of GENOMED, full professor at the University of Antwerp and affiliated professor at the Manipal University in India University of Antwerp*

Thursday, May 4, 2023; 3:00pm EST

More information coming soon!

Thursday, June 1, 2023; 3:00pm EST

Bone Turnover Markers

E. Michael Lewiecki, MD, *Osteoporosis Director, New Mexico Clinical Research and Osteoporosis Center, Inc.*

Thursday, July 6, 2023; 3:00pm EST

Pseudohypoparathyroidism

Emily L. Germain-Lee, MD, *Professor, Department of Pediatrics, University of Connecticut School of Medicine; Professor, Center for Regenerative Medicine & Skeletal Development, University of Connecticut School of Dental Medicine; Chief, Division of Pediatric Endocrinology & Diabetes, Connecticut Children's/UCConn School of Medicine; Director, Center for Rare Bone Disorders (includes Albright & OI Centers), University of Connecticut School of Medicine*

Understanding Clinical Trials

Supporting research is an important part of the OI Foundation's mission. There are five OI studies currently recruiting participants from the OI community. These studies focus on topics such as mental health, antibody treatment, and dental aligners. If you are interested in learning about or participating in these studies, please visit www.oif.org/currentstudies.

In the fact sheet below the OIF defines major terms and stages of clinical trials, explains the role of the FDA, and answers common questions.



What You Need to Know about Clinical Trials

Clinical trials are an essential part of developing new treatments (like medications, and vaccines) and test whether a specific treatment under consideration is effective and safe. Trials start with a small number of volunteer test subjects, and can grow to include thousands, or tens of thousands, of participants. Many clinical trials are *controlled*, *randomized*, and *double-blind*.

- **Controlled:** a clinical study with a group who receive the new treatment being studied are compared to a matching set of people who instead receive another treatment like a placebo, a harmless “fake” treatment (a placebo-controlled trial), a comparator drug (often a drug that is already approved for the condition under study), or different dosages of the study drug
- **Randomized:** participants are randomly assigned to either receive the new treatment or to be in the control group
- **Double-blind:** both the medical practitioners and the clinical study participants do not know who is receiving the new treatment and who is receiving a placebo (either the sponsor of the study knows the “key” to who is receiving the study drug or a designated person not directly involved in the study has the key)

Overall, these criteria help remove bias from a clinical study and ensure that results are accurate. In the US, the National Institute of Health (NIH) and the Food and Drug Administration (FDA) monitor clinical trials to ensure their safety and accuracy. While clinical studies of treatments for OI and other rare diseases have some differences, their basic structure is the same. All enroll volunteers at each phase, following protocols set out at the beginning of the study.

The phases of a study include:

- **Preclinical or Non-clinical testing:** scientists give the treatment to animals (like mice) to see if it produces a response and is safe
- **Phase 1:** a small group of human participants are examined; looking for safety, proper dosage amounts and to confirm it has some response in humans. Phase 1 may be done in healthy volunteers or in patients with the disease, or at-risk of the disease, under study.
- **Phase 2:** hundreds of people, further tests safety and if it stimulates response in different types of people
- **Phase 3:** thousands or tens of thousands of people; determine if vaccine or treatment is effective; a vaccine needs to be at least 50%-70% effective in protecting people. These are large enough to find relatively rare side effects that might be missed in earlier studies
- **Phase 4:** sometimes used after a treatment has been approved, this phase follows thousands of individuals and is used to find any long term effects of the treatment

Individuals with OI play an especially important role in the outcome of clinical trials because they have a rare condition. Compared to more common conditions, rare disease based clinical trials usually have far fewer participants that in the example listed above, making every volunteer even more valuable to the research process.

This article was prepared with assistance from Robert “Sandy” Sandhaus, MD, PhD, OIF Medical Advisory Council Member, September 2020.

Osteogenesis Imperfecta Foundation
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Serving the OI community with information and support since 1970

Clinical Trials and OI Questions

How do clinical trials focused on treating people with OI differ from trials focused on a more general population?

Since OI is a rare disease, it is difficult to find hundreds, let alone thousands, of participants. Therefore, rare disease clinical trials involve fewer participants at each phase. More information on clinical trials and how they differ for rare diseases can be found in Dr Adam Hartman's "[An Introduction to Clinical Trials](#)" presentation from July 23, 2020.

Are clinical trials safe to participate in?

While potential risks differ trial to trial and person to person, participating in clinical trials is safe, overall. Due to regulations in the US, volunteers are monitored at every stage of their involvement. Each participant in a clinical trial is informed about known and potential risks of the treatment being studied and will need to make an informed decision about whether those risks outweigh the benefits to themselves and the community when deciding whether to join the study.

How can I participate in clinical trials addressing OI?

The easiest way to learn more about current and upcoming clinical studies is to enroll in the [OI Registry](#). Once you enter your information into this registry, the OIF or researchers will have access to your deidentified information and may ask you to volunteer in a study. To find out about more information on specific clinical studies, you can also go to [Clinicaltrials.gov](#) and search for "osteogenesis imperfecta" in the "Condition or Disease" field. Here you will find an updated list of studies related to OI around the world.

Where can I learn more about clinical studies relating to OI?

The best way to hear more about potential clinical studies you can enroll is by enrolling at the OI Registry. More information can be found on the [OIF website](#). To find out about more information on specific clinical studies, you can also go to [Clinicaltrials.gov](#) and search for "osteogenesis imperfecta" in the "Condition or Disease" field. Within this search, you can filter studies by recruiting status, location, phase, and other criteria.

This article was prepared with assistance from Robert "Sandy" Sandhaus, MD, PhD, OIF Medical Advisory Council Member, September 2020.

Osteogenesis Imperfecta Foundation
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2023

New Year