This is not about Chopstix.
Chopstix is the tune budding piano players learn to play with two fingers. Right next to Chopstix in the repertoire of beginner piano music is Heart and Soul, a Hoagy Carmichael tune in which one person plays melody and a second person plays the bass line. More on Heart and Soul in a moment.

This issue of Breakthrough takes us through the recent important work going on at the OI Foundation. One of the key messages from the World Café (a long range planning exercise) was raising up issues around pain. Read about the Pain and OI project survey on page 4 of this issue. This survey invited people with OI to describe the pain they experience and the impact it has on their lives. An OI Echo session, an offshoot of the TeleEcho series, will cover pain in adults and children. And the Rare Bone Disease Alliance will hold a scientific symposium on pain in the fall.

An ongoing topic of interest in our community is the challenge of finding medical providers knowledgeable about OI in people who have transitioned out of pediatric care. In this issue you can learn about initiatives the OI Foundation is taking to provide exposure and training to more clinicians across the country, and even internationally. Look for stories on the OI Community, OI Clinic and Bone Health Town Hall meetings, the TeleEcho series, and more. I am grateful for our Medical Advisory Committee members for their role in this outreach.

There is more news around research and, of course, mutual support through conferences. I hope the conferences grab your attention! Heart and Soul is fun on the piano because it is a duet, something you play with a friend sitting next to you. OI Foundation events are returning to in-person gatherings where you can once again have a friend sitting next to you. Consider attending a regional conference in Phoenix, Arizona in September. In addition to getting the latest from the OI Foundation, you will be able to meet with friends old and new. Gathering and connecting with individuals and families with OI is the heart and soul of the OI community!

Ted Trahan

Members are the Heart of the OI Foundation

Without our members, the OI Foundation could not be the resource the OI community has come to rely on. From funding deserving research projects, to creating support groups, establishing regional and national conferences, and hosting fundraisers, we need your help! In this issue of Breakthrough, Jessica Sheridan shares why being a member of the OI Foundation is important to her and her family.

(continued on page 11)
The Importance of OI Research

Supporting research is an important part of the OI Foundation’s mission. Research helps advance diagnosis and treatment of osteogenesis imperfecta (OI) and helps medical professionals gain insight about the safety and effectiveness of drugs and other therapies.

A significant amount of OI research is coordinated by the Brittle Bone Disorders Consortium, part of the National Institutes of Health Rare Diseases Clinical Research Network. The BBDC allows physicians, researchers, and educators to focus on learning more about osteogenesis imperfecta and developing new and better treatments to improve the care of those living with OI. This program has sped up the pace of research and put useful information into practice much quicker than otherwise possible by using the skills of experienced researchers from many institutions, creating access to research centers closer to where people live, and expanding educational opportunities for healthcare providers.

You can learn more about the work of the BBDC through the OI Foundation’s Current State of OI Research video series. This series was created to highlight and describe the BBDC, including current studies and research related to OI. Some of the topics featured include Women’s Health and Pregnancy, Dental Health, Mental Health, New Medical Treatments, and an overview of the OI Longitudinal Study. To watch the videos in this series, please visit the OIF website or the OI Foundation’s YouTube channel at www.youtube.com/@OIFoundation.

How Can I Participate in OI Research?

Often, the success of clinical studies of a rare disorder like OI depends on getting enough people to participate in the study so the results are meaningful. Your participation is critical to move OI research forward.

Visit www.oif.org/currentstudies. The OI Foundation strives to keep OI community members updated about all current and ongoing OI studies. The OIF’s Current OI Studies and Publications webpage features studies that are currently recruiting participants.

Join the OI Registry. Individuals with OI (18 and older) and parents of children with OI are encouraged to join the OI Registry. The OI Registry is a database of individuals with OI who are interested in participating in OI research. Once you are enrolled, you will begin to receive information on upcoming studies and online surveys about OI. Your participation in these studies helps move OI research forward. To find out more please visit www.oif.org/oiregistry.

Learn More about participating in a clinical trial. The OI Foundation’s What You Need to Know about Clinical Trials factsheet defines major terms and stages of clinical trials, explains the role of the FDA, and answers common questions. This and other OIF factsheets are available on the OIF website’s Information Center at www.oif.org/factsheets.

Visit the OIF’s Current Studies and Publications Webpage

The OIF’s Current OI Studies and Publications webpage features studies that are currently recruiting participants, as well as recent publications of OI research. This page also includes a spreadsheet, by Riley Johnson B.S. (Research Assistant, Oregon Health and Science University), created as a resource for people interested in the medical science supporting osteogenesis imperfecta (OI) in adults. This spreadsheet includes more than 200 articles published in the last 40+ years. Learn more at www.oif.org/currentstudies.

Thank you, Dr. Francis Glorieux, for more than 10 years of service as the Chair of the OIF’s Medical Advisory Council!

As Chairman, Dr. Glorieux led the OIF Medical Advisory Council (MAC) and volunteered his time and expertise to review OI Foundation publications, advise the OI Foundation on new research, speak at OIF events and conferences, and answer questions from the OI community. We are so thankful to Dr. Glorieux for his leadership and his commitment to children and adults living with OI. We are pleased to share that Dr. Glorieux will continue to serve as a member of the OIF’s Medical Advisory Council and the OIF’s Board of Directors. The OIF continues to be so grateful to Dr. Glorieux for all he does for the OIF and the OI community. Learn more about Dr. Glorieux at www.oif.org/glorieux.
Preventing Adults with Osteogenesis Imperfecta to Engage in Research on Access and Quality of Care for Their Rare Disease

The OI Foundation’s 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), has concluded its phase of surveys, interviews, and listening sessions, as announced in previous Breakthrough issues. Surveys were sent out to both patient advocacy group leaders and pediatric clinic leaders, interviews were done with leaders of patient advocacy organizations and various medical professionals, and listening sessions were held with members of the OI community including patients, parents, and caregivers. Topics explored in the ventures of this phase included 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 health care across the age span.

These methods have helped us to identify major gaps in care transitions and adult care for individuals with OI, including the following:

- Patients with osteogenesis imperfecta are not given instructions on what doctors and/or specialties they need to see after they reached adulthood and were dropped by their pediatric care team; many participants were not given referrals.
- Patients are often dropped by adult care physicians before their new patient appointments once those physicians learn they have OI.
- Emergency rooms are often avoided by patients with OI when they experience fractures, as the expense is high while the knowledge around OI is lacking.
- Patients often feel overwhelmed when it comes to consistently having to educate the physicians they see in their adult years on OI.
- Lack of adequate insurance becomes a huge roadblock for patients transitioning to adult care.
- Finding access to general providers that take disabilities and rare diseases into account when dealing with more “common” illnesses is a problem. Patients feel as though they need to see an expert, even for basic viruses.
- Adults with OI should see multiple care providers in various specialties including audiology, cardiology, endocrinology, and pulmonology. Patients were not advised to put together these care teams as pediatric patients.

Other notable common themes from the listening sessions and interviews include:

- Patients agree that while they should take charge of their own care, it should be the responsibility of their pediatric care team to prepare them for the realities of adult care.
- As a parent, it is important to have your children with OI become more involved in their own care from an early age, so they know how to properly advocate for themselves as adults (i.e., speaking to the doctor themselves about how they are feeling, setting up their own appointments as teens, getting involved in their treatment options, etc.).
- In addition to finding medical care, finding employment and affordable housing is hard for adult OI patients. In turn, this appears to affect both the mental and medical health of those patients.
- OI clinics need to begin networking with local providers to spread awareness and information about patients with OI.
- The next generation of providers need to be educated on OI as the experts currently in the field get older. There is some concern from leaders of patient advocacy groups that they aren't seeing this next generation of rare disease experts come up.
- Participants from the listening sessions have a generally optimistic outlook on the future of OI care for the next generation, as long as the current gaps in care are properly addressed.

Now that we have completed the surveys, interviews, and listening sessions, the strategic planning will begin. This project will continue to run through October of this year, so stay tuned for more updates.

We want to hear from you!

Do you believe any major gaps in care transition were missed? If you have anything to add, please reach out to Stacie Connors at sconnors@oif.org.
Understanding Pain and OI

In the past few years, researchers have been putting more focus on understanding how pain impacts people with OI. While pain has a large impact on people’s lives, it is unclear how it specifically impacts aspects of a person’s well-being and health. One current group that is investigating these questions and others is the Brittle Bone Disease Consortium (BBDC). Through the BBDC, OI researchers have been conducting interviews with individuals with OI about their mental health and their experience of pain. These interviews are part of a study aiming to better understand the mental health concerns for people with OI. The OIF is a proud partner of the BBDC, and has interviewed the leaders of this study previously. To view this recording, please visit the OI Foundation YouTube channel.

Another group looking into these questions is the Pain and OI Project, a group co-led by the OI Foundation and the Osteogenesis Imperfecta Federation of Europe (OIFE). This group consists of international OI advocacy leaders, researchers, and medical professionals from 12 different countries. One project they undertook was to create a survey where all adults with OI could share information about their pain and how it impacted them. This survey gives a snapshot of a group of adults with OI, and may help contextualize an individual’s experience and suggest future areas of study for more research.

The Pain and OI survey was open from January 23, 2023, to April 1, 2023, and collected 1,086 responses from 49 countries, with 55% of total responses coming from the US. Information was gathered about people’s demographics, OI type, mobility, fractures, and orthopedic surgeries. Participants were also asked about the different types of pain they experience, pain location and duration, and how they treat their pain. Below are some major initial findings from the survey. The OIF hopes to present a more thorough analysis later.

As with any anonymous community survey, the findings should not be taken as a fact, and may not necessarily be scientifically accurate. Some, but not necessarily all, factors that may have impacted the accuracy of the results include:

- The OIF cannot verify if the survey responses were truthful or medically accurate.
- The 1086 people who completed the survey may or may not be a good representative sample of the entire OI population. Certain groups may be over or underrepresented in the results.
- The survey was only provided in English, and some attendees may have had language difficulties.
- The survey was only distributed online, and individuals needed internet access to complete it.

Finding #1: Many people with OI appear to experience lots of chronic pain.

40% of responses said they had acute pain (pain caused by injury, surgery, illness, trauma, or painful medical procedures) in the past four weeks, while 78% of responses indicated they were currently experiencing chronic pain (pain that had lasted longer than three months). 87% of people indicated that they had experienced pain in the last 12 months, and 60% had experienced pain every day in the last 4 weeks. These survey results may indicate that many people with OI have long lasting pain and nearly constant pain.
Finding #2: Back pain is common for people who completed the survey.

The Pain and OI Survey also gathered information on where on the body people felt pain. Question 19 asked, “If you are experiencing pain right now, where are you experiencing it on your body? (Check all that apply).” 19 body parts were listed along with a space to input other options, and 800 people answered the question. The first and fifth most common answers were the spine and neck, with 55% and 35% of responses, respectively. While they felt pain in other areas as well, four out of the top five most common responses directly or indirectly could contribute to back or torso-related pain.

Finding #3: Pain impacts many parts of people’s lives including career and social life decisions.

Regardless of its severity, pain can impact quality of life. The survey asked participants to rate how pain impacted different aspects of their lives, using the choices of “Not impacted, very mildly impacted, mildly impacted, moderately impacted, severely impacted, I don’t know, and prefer not to say”. In the 14 life aspects that individuals rated, pain had at least some impact on all of them. In particular, the aspects with the highest percentage of people indicating that pain had “severely impacted” were career choices with 34%, the type of job they could do with 30%, and the types of leisure activities they could participate in at 30% as well. While pain had a large impact on physical health, people’s social and financial lives were not spared.

What’s Next?

Pain and OI, like our understanding of OI itself, is complex. We hope that the results from this survey can help contextualize the pain that OI community members feel, and that it can help drive further research.
Living With Osteogenesis Imperfecta: Understanding Experiences Based on Community Insight and Evidence Survey, the IMPACT Survey

What is the IMPACT Survey?
In 2021, the Osteogenesis Imperfecta Foundation (OIF), the Osteogenesis Imperfecta Federation Europe (OIFE), and Mereo BioPharma collaborated closely to launch the IMPACT Survey. The goal of this project was to capture and quantify the real impact OI has on the lives of people with OI, their families, and caregivers. We were thrilled that more than 2,200 OI community members from 66 countries participated in this survey!

The survey was based on evidence gaps around:
- **Clinical impact** of OI on affected individuals
- **Humanistic impact** on individuals, their families, and caregivers
- **Economic impact** on individuals and wider society

The IMPACT survey is now closed. Thank you to all participants who made this possible.

What Were the Target Groups?
The survey was developed for the following primary target groups:
- Adults with OI (over 17 years old)
- Parents (without OI) of children with OI
- Parents (with OI) of children with OI
- Adolescents with OI (12 – 17 years old)

In addition to the target groups above, there were also a substantial number of responses from:
- Parents of adults with OI
- Close relatives of people with OI

There were different questions based on which target group you belong to. Adults with OI who had children with OI, could answer both on behalf of themselves and their children.

What Will the Data be Used For and How is It Stored?
The IMPACT Survey provides a large global dataset describing the experiences of the OI community. The results of this survey are being used to improve healthcare services and potential future treatments for OI.

The data was gathered on a confidential and anonymous basis, with Non-Personally Identifiable Information (Non-PII). It is securely stored by Wickenstones according to Data Protection Regulations. The data is now being analyzed according to the plan developed by the Steering Committee, to prepare four proposed central publications on the impact of OI in peer-reviewed, scientific journals.
What Data has Been Published?
What are the Findings?

So far, the first article “The patient clinical journey and socioeconomic impact of osteogenesis imperfecta: a systematic scoping review” was published in Orphanet Journal of Rare Diseases in February 2023. The purpose of this review was to capture and quantify the published evidence relating specifically to the clinical, humanistic, and economic impact of OI on individuals, their families, and society. The review suggests that there is limited data regarding health concerns beyond bone health and how these concerns may impact health-related quality of life, in particular, that of adult men and other family members. The four remaining articles will be published in 2023 and 2024.

The graphic below shares the most common health conditions in adults with OI (of the past 12-months prior to answering) based on the responses from participants. Pain, fatigue, and soft tissue are the most common symptoms, while fractures are in 13th place for the adult population.

Learn more about the IMPACT Survey and the findings from this project at [www.oif.org/impactsurvey](http://www.oif.org/impactsurvey).
Each year, leading scientists and medical professionals attend the OI Foundation Scientific Meeting, a two-day event where attendees collaborate and share research. OIF Medical Advisory Council (MAC) members, researchers in the Brittle Bone Disorders Consortium (BBDC), and other important researchers and clinicians are invited to attend. This year’s meeting, held on April 13-14, 2023 was chaired by Dr. Kenneth Kozloff (Professor of Orthopaedic Surgery at the University of Michigan). The meeting, titled Transforming OI Research Together, featured over 30 presentations on a wide variety of topics including Energy Metabolism, Growth, OI Organizations, and Emerging Therapies.

To help better understand the OIF Science Meeting and its importance, the OI Foundation spoke with Dr. Frank Rauch, a member of the OIF’s MAC and longtime Scientific Meeting attendee and presenter. In an interview with OIF staff, Dr. Rauch explained the structure of the Meeting, how it helps push research forward, and what topics he found particularly interesting from this year. The following transcript has been edited and shortened for clarity. To watch the full interview, please visit the OIF’s YouTube Channel.

What is the OIF Science Meeting, and can you give us a brief overview of what type of people are attending it?

**Dr. Frank Rauch:** The OIF Scientific Meeting happens every year and it’s by invitation for researchers who are interested and active in the field of researching OI. It is a very important meeting because it brings together people that work in the same field, so the exchange of ideas and discussions is very important for bringing ideas forward. I think many of the advances over the past 20 years have started at one of these Scientific Meetings. I think this is really a model for many other diseases, where such things may not happen. I think they can copy that from us. So, the way it works is usually, I think, 70-80 people are invited and then the meeting is over one and half days with about 25 presentations. People show the latest results and then there is time for discussion among the people who do similar things or have similar experiences. We really, really flesh things out.

Can you tell us a little bit about why this meeting happens every year? Obviously new things are getting brought up, but are there some topics or large themes that are getting brought up again and again?

It’s dependent on what problems people face. And of course, bones and fractures are a topic that is always there, all the time. Also, depending on who is organizing the Meeting, or what the focus is for that year, sometimes the focus is on orthopedic issues. Sometimes it’s more basic science issues that are discussed.

Over the past few years, we have had many presentations on things other than bones, so these broader problems that are associated with OI have been more widely discussed. And I think these meetings also set the agenda for what is discussed outside of these meetings, because I think all the opinion leaders are there. **This is really a trendsetter meeting that determines what people are really interested in, in the field of OI research.**

You brought up the importance of covering basic research versus clinical research. Can you explain what that difference is?

So the easy thing to explain is clinical research because most people who have come into direct contact with research have participated in clinical research. That’s basically collecting information about people with a condition or people that are in the hospital or wherever you study them. So clinical research is whenever research includes people. Basic research is any research that does not include people. That is just purely on molecules or on cells, but in many cases it’s about animal models. So in OI, we have animals that are similar in appearance to OI in humans. A lot of the basic science in OI is dealing with mice.

Can I ask why we study mice?

Good question. You know, I’m a clinician myself, so I have the same question. Why is everyone talking about mice and not something else? **Well mice are practical, because they are small and breed very quickly.** So, if you want fast results, it’s an advantage to have mice because eight weeks after they are born, they can already reproduce.

**Also, the other practical aspect is that in mice you can introduce genetic changes very easily**, and that’s much more difficult in other types of animals. People have been able to introduce variants that they have seen in people with OI, and they made the same variants in
the mice. Then they were able to see the effect of these variants in the mouse models. We have five or six different mice that have some kind of OI variant in their genes, and they are reflecting, to some extent at least, the problems that we see in people.

These models are useful, because you can first test any new treatments or new ideas on a mouse model before you even consider using the same approach in people.

It’s determining the safety or viability aspect of the research, it sounds like.

One thing is safety. But also, you know, does it even work? Is the idea useful? If something has no effect in mice, it probably will not be developed further. And the other thing is safety; it might have side effects. So it’s this filtering step of developing new approaches for treatment.

So you had mentioned that this meeting is a place where thought leaders are getting together to discuss new trends and updates together. What role is the OIF playing in this meeting? Because the OIF itself is a patient-advocacy group.

The OIF is a key player in this field, of course. For this meeting, it’s organized by the OIF, so all the logistics are done by OIF and I think OIF is also great at providing a friendly environment that is not on anyone’s particular field, so it’s like a neutral ground. It’s not like one researcher invites everyone else. Everyone is coming to the OIF. And it’s not a trivial task to invite so many people and deal with all the logistical issues around it. And so that’s very nice, that’s the first, they have a lot of experience with organizing events.

I want to ask why this meeting isn’t recorded or more publicly available? The OIF prides itself on trying to publish or amplify the work that researchers are doing. But with this meeting, we don’t invite the public or record it.

I think if you want a meeting where the latest ideas are discussed, you don’t want everything to be public, because the ideas may change next year and then you have it recorded on YouTube forever. It’s difficult to change ideas if everything is already documented. The other thing, of course, is that you have no control where these ideas go that you mentioned in a meeting like this, and people may scoop you. Whereas, if you keep it in the room where the meeting takes place, you can trust the people that are there, and you can really talk more freely. I think in recorded meetings, typically people talk about the things that are already public. The things that we talk about in the OIF Scientific Meeting will be public in two or three years, so it gives us a head start when you take part in this.

This year’s Scientific Meeting just happened. Can you tell us a little bit about it? Who hosted this year and what were some of the large takeaways from 2023?

So this year the meeting chair was Dr. Kozloff from University of Michigan. And every year the OIF invites somebody new to make the program. And he did a very nice job in finding new topics that have been discussed less in the past.

One topic that was a whole new session was Energy Metabolism in OI. There are many studies that point towards some problems with energy metabolism, which includes things like heat production in the body, insulin secretion, glucose metabolism, and more. So, there are many things that are related to this topic and there are some studies on individual issues, but I’ve never seen
a whole session on this where all the people working on this topic were together in one group. I think that was very interesting. In people with OI, this is a topic that has not really been addressed much in our clinical practice. So that’s still in the discovery phase.

The next topic that I have found very interesting for long time already, is growth and OI. Which is a bit of a mystery until now. Why do many people with OI have short stature? Why do kids with severe OI not grow well? There were presentations that shed some light on these questions. That was a first for me, so that was also very interesting. We also heard about some new mouse models of OI, so people developed new mouse models that better reflect the issues that we see in people with OI. So that was like the program of the first day. And then the second day, was about the clinical projects that are going on. So, an overview of the important ongoing clinical projects, like the Brittle Bone Disease Consortium (BBDC) is the one big study across North America.

And finally, there were presentations of the different companies that developed medications for OI, and this, I think, was also a new part of the program. There are enough companies that are interested in OI right now to have a whole session on them. This is quite an encouraging development, that so many companies also have an interest in this area.

It is very exciting to hear that there’s more industry interest and participation in this field of OI research! Looking ahead now to future years, are there topics that you are most interested or excited about?

Research is unpredictable. Really good research should surprise everyone. So, I hope to be pleasantly surprised in the future. But the things that I’m interested in are usually the topics that I don’t know much about or that nobody knows much about. I think this growth issue has big potential of becoming more important. Slow growth is such an obvious and important part of OI that we must figure out why that happens precisely and then develop treatments.

Another thing that has not been discussed much are gastrointestinal issues. We had some discussions on that in mouse models. I just often hear this from patients, but nobody has really studied this in any detail, and nobody knows exactly where it’s coming from. So, I think people are starting to get interested in this aspect. There are certainly new drugs that will be developed and tested in the next coming years. So, I think these are new topics that we are going to see in these meetings.

Thank you, Dr. Rauch, for sharing your insight on the OIF Scientific Meeting. It’s incredible to bring together so many top researchers to move OI research forward.
OIF member Jessica Sheridan shared why being a member of the OI Foundation is so important to her and her family in the interview below.

**What is your relationship with OI?**

My daughter was diagnosed with OI in 2012, 6 weeks after she was born. It was spontaneous, rather than genetic, and at that time we knew no one with OI and had no prior knowledge of what it was. We were put in contact with the OIF, which was a wonderful experience because the resources we were able to utilize and the people we met (both staff and other families) helped us cope and navigate the diagnosis as first-time parents.

I wanted to learn more and more about how best to help my child but, in the meantime, I found this network of people who devoted themselves to spread awareness about this rare disease and worked continuously to develop creative ways to raise funds to help the community.

**Why is membership in the OI Foundation important to you?**

I am constantly amazed by all of the hard work and dedication that goes into all of the events that the OIF community plans, and all of the ways the OIF helps the OI community. At the heart of it all is a dedicated network of people working together to help support those living with OI, which is a cause near to my heart, of course. I have such gratitude for all of the people who care about my daughter. Not to mention, the community is so positive. When you have a loved one with OI it can be difficult, but knowing that there are so many people working together to help and who care is amazing.

**Have you ever attended an OIF meeting/conference/event?**

I have yet to attend a conference, but when my daughter was young we planned golf outings in Michigan and Illinois for a few years, which will always hold very special memories to me.

**Is there any advice you would give to a new family/member of the OI Community?**

When things in life seem unexpected, it can feel very confusing and scary. However, the most difficult parts of life also happen to be the most wonderful opportunities to experience immense love, compassion, and kindness from others. There are so many people to lean on in this community, and people who care and want to help you on this journey we are all traveling together. Don’t feel that you are alone. Reach out to others, don’t be afraid to ask for help and use invaluable resources such as the OIF to help assist you. Go to an event if you can and meet people, or plan an event and watch people come out in droves to show you love and support. There are so many of us out here who are now part of your tribe!

**What upcoming OIF program are you most looking forward to and why?**

I love hearing about all of the many ways people all over the country are continuously working together to spread awareness. I love the Bone China Tea. I love reading about new grant opportunities and new medical research. It gives me a lot of hope for the future.

The future of the OI Foundation depends on you! We count on the support of our members to help fund research, provide information and support, develop new resources, expand support groups and increase public and professional awareness about OI. Without you, the vision for the OI Foundation once had by a group of parents in 1970 would not have become the resource it is today. From funding deserving research projects, to creating support groups, establishing regional and national conferences, and hosting fundraisers, you’ve been with us through it all.

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**PLEASE CONSIDER BECOMING A MEMBER OF THE OI FOUNDATION TODAY!**

**Membership Levels**

- **Bronze** $36 per year
- **Silver** $100 per year
- **Gold** $365 per year

This membership comes with an Unbreakable Spirit® cinch sack bag and a complimentary ticket to an OIF fundraising event or regional conference of your choice.

**1970 Society** $1,000 per year

This membership comes with an Unbreakable Spirit® drawstring bag, complimentary ticket to an OIF fundraising event or regional conference and a gold 1970 society lapel pin.

All members will receive an annual subscription to the OI Foundation’s printed newsletter, “Breakthrough”, invitation to an exclusive program update meeting and voting privileges for Board Member elections.

Please consider becoming a member of the OI Foundation by filling out the attached envelope or visiting [www.oif.org/membership](http://www.oif.org/membership).
OI Clinic Spotlight: Children’s Healthcare of Atlanta

The OI Foundation works closely with nearly 60 multidisciplinary OI Clinics across the continent to provide timely and accurate information about the range of available services to OI community members. The complete OI Clinic Directory can be found at [www.oif.org/ClinicDirectory](http://www.oif.org/ClinicDirectory).

To provide an overview of the background, mission, and services of these centers, the OIF spotlights new and existing OI clinics serving pediatric and adult patients in each issue of Breakthrough. In this issue, we are excited to introduce the **OI Clinic at Children’s Healthcare of Atlanta** in Atlanta, Georgia.

**Children’s Healthcare of Atlanta: OI Clinic**

Children’s Healthcare of Atlanta (CHOA) is Georgia’s leading pediatric healthcare provider. Consistently ranked among the top children’s hospitals by U.S. News & World Report, the CHOA has a team dedicated to treating children with OI and it is one of the highest-volume pediatric hospitals in the country treating OI patients.

Starting June 2023, CHOA will run a multidisciplinary OI Clinic on the second Thursday and Friday of every month, with visits spread over the two days. The CHOA recognizes that many of their OI families travel to receive care. Therefore, CHOA wants to provide a range of services to minimize families’ travel time and maximize their care across multiple disciplines. To help families traveling to the clinic, CHOA will have housing options available at two Ronald McDonald houses, which can also provide transportation to the hospital for services if needed.

The OI Clinic’s primary goal is to provide patients with a coordinated care approach through a multidisciplinary clinic to better manage the OI diagnosis with a holistic treatment plan. Patients being seen at the OI Clinic can receive imaging, pulmonary function tests, dental exams, hearing screenings, physical/occupation therapy, endocrinology, and orthopedic exams.

To contact Children’s Healthcare of Atlanta about their services, please call 404-785-5438 or email at [oiclinic@choa.org](mailto:oiclinic@choa.org).

**Camp Wishbone Family Weekend**

Camp Wishbone is a weekend getaway held at Camp Twin Lakes in Winder, Georgia. Campers with osteogenesis imperfecta (OI) and their immediate family members are invited to enjoy camp activities with others who share similar experiences.

During the weekend, families have the opportunity to participate in a variety of camp activities including canoeing and kayaking, fishing, horseback riding, ceramics, biking, arts and crafts, archery, and climbing. To help support caregivers, Camp Wishbone offers parent only sessions throughout the weekend. Caregiver sessions may include a guest speaker or a
special activity. Family weekend can be a great time to introduce camp to new campers with the support of family members.

Made possible through the generous support of our donors, Camp Wishbone is offered at no cost to families. CHOAs camps rely on donor support and every dollar counts.

Camp Wishbone reserves space on a first-come basis. Since space is limited, only parents or guardians and siblings of our campers may attend.

CHOAs medical team will review all applications before granting final approval.

When: September 15 – September 17, 2023

For more information, please contact Catherine Shields at Catherine.shields@choa.org or call 404-785-KIDS (5437).

Introducing the Newest Michael Geisman Fellowship Grant Recipient

The OI Foundation is excited to introduce the latest Michael Geisman Fellowship Grant recipient for the 2022 cycle: Giulia Montagna, PhD.

The Michael Geisman Fellowship Grant program awards funding up to $50,000 per year, for two years, to postdoctoral trainees who are currently working on projects with clear relevance to osteogenesis imperfecta, or who have projects that will enable them to develop expertise in OI research.

Giulia Montagna is a postdoctoral research fellow at Boston Children’s Hospital, affiliated with Harvard Medical School. She holds a bachelor’s degree in Biomolecular Biotechnology, a master’s degree in molecular biology and Genetics, and a Doctor of Philosophy in Health Technologies, Bioengineering, and Bioinformatics from the University of Pavia in Italy. Her research project, A Novel Approach for Studying Pain in an Improved Genetic Mouse Model of OI, focuses on chronic pain in patients with OI by using a conditional genetic mouse model to understand the source of that pain, and whether it originates directly from bone fractures or other concurrent mechanisms such as microfractures, tendon and ligament injuries, periosteal tears, and so on. In measuring animal movements, Dr. Montagna found that five-week-old mice with the Aga2 mutation moved significantly less than their wild-type littermates, but also had an average of 7 fractures. It was not possible to determine if pain in those mice was caused by those fractures or by other factors preceding them. Her approach includes inducing the expression of the Aga2 OI allele and assessing mobility and indicators of pain before and after the mice begin to experience fracture. If signs of pain are observed before evidence of fracture or deformity, the source of that pain can proceed to be better defined. Therapies can then be tested to alleviate that pain.

In her spare time, Dr. Montagna enjoys playing sports such as volleyball, skiing, and running. She enjoys traveling with friends whenever there is time and possibility to arrange group trips.

We look forward to sharing more about Dr. Montagna’s important work!

“I decided to start this project because of my passion for bone biology and my background in molecular biology. This was the perfect combination of these two things. Once I started, it became more and more important to succeed in the project because this might have almost direct repercussions on the patients that have this condition. The idea to help patients with a rare condition to better cope with the pain that condition causes is very motivating to me.”

– Dr. Giulia Montagna
OIF Conference Program

The OI Foundation Conference program brings together OI community members to learn from OI experts and connect with fellow OI families.

OIF National Conference

The OIF’s biennial National Conference is the largest informational and social event for families and individuals living with osteogenesis imperfecta, bringing together more than 600 members of the OI community. Over the course of three days, OI families attend medical information sessions led by experts in OI from around the world and participate in social activities that help build support systems that last long after the conference has ended.

The OI Foundation’s Medical Consultations Day is one of the most popular programs offered at the OIF National Conference. Medical consultations offer families and adults living with OI the opportunity to have a face-to-face meeting with a physician or physical therapist who has extensive experience with OI. It is the chance to get questions answered, to have a test or diagnostic letter explained, or to get a second opinion about a treatment.

Since 2020, OIF National Conferences have been held virtually each year. These virtual events provided live online versions of some of the most popular informational and social sessions at the biennial OIF National Conference. To watch recordings from past OIF Virtual Conference sessions, visit the OIF’s YouTube channel.

OIF Regional Conference

The OIF’s Regional Conference program was created to bring information and community to families living with OI across the country. Regional Conferences are one-day educational sessions that cover topics essential to understanding OI held in cities across the country. Local families come together to hear from OI experts, meet medical professionals, and connect with OI community members in their area.

We hope you will join us at an upcoming OIF Conference!

Upcoming OIF Conferences:

OIF Regional Conference: Phoenix, AZ
September 9, 2023
Phoenix Children’s Hospital

OIF National Conference 2024: Omaha, NE
July 19-21, 2024
Hilton Omaha
Connect with OI Community Members in Your Area Through OI Support Groups

One of the cornerstones of the OI Foundation’s mission is to improve the quality of life for people living with OI and their family members through mutual support. The OI Foundation recognizes the value of creating a space where members of the OI community can share their personal experiences, and both receive and provide support. One of the OIF’s resources for connecting individuals with OI and OI families is the OIF Support Group program.

With the return of in-person events, the OIF is in the process of revitalizing and revamping the OIF Support Group program! OIF Support Groups will consist of five regional (Northeast, Southeast, Southwest, Midwest and West) and various affinity groups. Regional support groups will be held virtually and have the option for in-person meetings once or twice a year, depending on the group’s desires. Affinity groups will be held virtually throughout the year.

Support Group Meetings are, first and foremost, a social gathering to bring local OI community members together, though speakers may be scheduled from time to time to provide information on topics of interest to the group. Some groups choose to organize local fundraisers and raise awareness about OI in their communities.

Get involved! OI community members are encouraged to get involved in the OIF Support Group program! The OIF is currently seeking Support Group Leaders (community members who organize Support Group meetings), and State-based Contacts (community members available to offer support, answer questions and direct people with OI and their families to resources). All Support Group Leaders and State-based Contacts will receive training from the OI Foundation.

If you are interested in being a regional or affinity Support Group Leader or becoming a State-based Contact for your state, please contact Kenna Coleman at kcoleman@oif.org for more information.

Nominate an OIF Board Member!

The OI Foundation is always seeking nominations for new members of the OIF Board of Directors. The Board Development Committee looks for nominees with qualities such as a personal connection to OI and experience in areas including accounting, finance, fundraising, organizational management, public relations, strategic planning, business and/or law.

If you or someone you know is committed to achieving the mission of the OI Foundation please contact the OIF at bonelink@oif.org.
Did you know that 20% of the OI Foundation’s annual operating budget comes from volunteer fundraising events? The OI Foundation would not be able to provide the beneficial programs and services we are known for without the support of events such as the Fine Wines Strong Bones galas, golf outings, walk-n-wheels, and countless personal fundraisers. Whether they are sipping on fine wines or trying to make a birdie on the golf course, OI community members have converted activities they are passionate about into ways to raise money for the Osteogenesis Imperfecta Foundation.

Over twenty years ago, a group of OI Foundation volunteers hosted the first Fine Wines Strong Bones event to educate the community about osteogenesis imperfecta (OI). Over time, the event has grown into a vibrant, annual affair celebrating the Unbreakable Spirit® of the OI community. The flagship Fine Wines Strong Bones event has inspired similar food and wine gatherings in Boston, New York, Houston, Chicago, Tampa, and Naples.

The original Fine Wines Strong Bones DC event has undergone many forms in its 23-year history – starting as a wine tasting event after work, then becoming a Saturday evening black-tie optional gala, and even going virtual for two years. This year, the event was transformed again to meet the needs of the local OI community members who form the host committee. Tee Time at Fine Wines Strong Bones DC was held at Top Golf National Harbor on May 6, 2023. The indoor/outdoor space was the perfect accommodation as we held our first in-person event in the DC Metro Area since 2020. The 100 attendees participated in silent auctions, raffles, some golfing, and the fund-the-mission paddle raise to support the Jamie Kendall Fund for OI Adult Health (in memory of former OI Board President and original Fine Wines Strong Bones DC Committee Member, Jamie Kendall).

In March 2023, Teresa & Ken Gudek and Jane & Jim Early, held the 8th annual Fine Wines Naples Reception and raised $95,000 to support the programs and services of the OI Foundation. It was an incredible night, with an exceedingly generous network of people who attended.

OL Foundation Board Vice President Jim Early shared, “I love the opportunity to fundraise. My family, in partnership with the Gudek family, has had tremendous joy in hosting both the Fine Wines Naples Reception and the Matthew Smith Memorial Golf Tournament (formerly the OI Golf Classic) in New Hampshire. It’s very satisfying to see the support we get from those who contribute so generously, but more important for me, it’s an outlet from which I can share my passion for all that needs to be done to improve lives for those in the OI Community.”

Summertime brings a variety of golf outing fundraisers taking place across the U.S. including both the Riley’s Gathering Golf Outing and Miracle Michael Golf Outing in Chicago in July; The Matthew Smith Memorial Golf Outing in New Hampshire in August; and the 2nd Annual Unbreakable Spirit® Golf Tournament in Pennsylvania in September; Visit the OIF’s Upcoming Event calendar online (www.oif.org/events) to keep up to date with all fundraising events that are open to the public.

If you are interested in fundraising but the idea of hosting an event intimidates you, you’re not alone! Bone China Tea (BCT) is an annual fundraising campaign of the OI Foundation and was a virtual event (before Zoom meetings even existed!). Hosts contribute to OI awareness, support, and research efforts by inviting their friends and family to enjoy a cup of tea in the comfort of their own home and take the opportunity to donate a gift to the OI Foundation with the money they may have saved by not going out to an in-person event. Chair of the Bone China Tea program, Susie Wilson, hosted her first Bone China Tea fundraiser with her mom in 1997 and has chaired the event solo since 2016. Susie explains, “what is great about this fundraiser is you don’t have to get all dressed up, get a babysitter or travel a great distance to attend. You can participate in this fundraiser right from your own home at your kitchen table. It is a fun simple way to update your family, friends and co-workers about you, or your loved one with OI and how they are doing. Another wonderful way to help bring awareness to those that may not know much about OI.”

Our donors and members are the backbone of the OI Foundation! Join fellow OI community members in supporting the OI Foundation by turning something you are passionate about into an OIF fundraiser. Whether you want to donate the money raised at a yard sale, run a marathon, host a Blue Jeans for Better Bones Day, or hold a family fun day, the OIF fundraising staff is here to help! Please contact Cait at cfarren@oif.org if you are thinking about holding an event to raise money for the OI Foundation, or if you would like to be on a committee that plans a fundraiser.

Thank you! Your support helps fund OI research and expands our reach to ensure the OIF’s programs and services are accessible to every person with OI.
Introducing OIF Medical Advisory Council Chair, Dr. Cathleen L. Raggio

We are pleased to announce that Dr. Cathleen L. Raggio will serve as the chair of the OI Foundation Medical Advisory Council (MAC) beginning July 1, 2023. Dr. Raggio is a pediatric orthopedic and spine surgeon who specializes in skeletal dysplasias (i.e., osteogenesis imperfecta), and scoliosis.

Dr. Raggio is affiliated with the Hospital for Special Surgery. She graduated from Weill Medical College of Cornell University and completed her orthopedic training at the Hospital for Special Surgery and the Alfred I DuPont Hospital for Children. She has been recognized for her outstanding work through numerous awards including the Castle Connolly 2023 Exceptional Women in Medicine distinction, awarded to those with exceptional leadership, expertise, and dedication in their respective fields. She conducts both clinical and basic science research as a clinician scientist.

As a longtime OI Foundation MAC member, Dr. Raggio has volunteered her time and expertise to review OI Foundation publications, and to advise the OI Foundation’s CEO, staff, and Board of Directors on the implications of new research discoveries and new medicines or treatments related to OI. She has been featured as an OI expert speaker at OIF National and Regional Conferences, Support Groups, Scientific Meetings, and more.

“Being a MAC member is a great honor and responsibility. It is the best example of team work and collaboration for a common purpose (advancement of care for the OI community) that I can imagine.”

– Dr. Cathleen Raggio

Thank you, Dr. Raggio, for all of your contributions to the OI Foundation and OI community!

Advocating for the OI Community

One of the goals of the OI Foundation is to support fair access to patient care. The OIF collaborates with fellow patient advocacy groups through the Haystack Project and The Global Advocacy Alliance to advocate for OI community members.

The Haystack Project

The OIF is a member of the Haystack Project, a collective of rare and ultra-rare (20,000 or fewer US patients) disease patient advocacy organizations that work together to amplify the voices of rare and ultra-rare in issues impacting availability of and access to treatment. They strive to amplify the patient and caregiver voice in states where the unmet need is high and treatment delays and inadequacies can be catastrophic.

In 2022, the Heart Act was passed, which advocated for patients and experts to have an increased role during the FDA review process for treatments for rare diseases.

To find out more about the Haystack Project, visit www.haystackproject.org.

The Global Advocacy Alliance by Nonprofit Global Genes

The OIF is a member of the Global Advocacy Alliance, a global community of non-profit organizations and support groups, whose mission is to provide information, resources and connections to all communities affected by rare diseases. Together, members can connect, empower, and inspire the rare disease community.

Learn more at www.globalgenes.org.
Thank you to our Members!

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www.oif.org/conference