

# Melorheostosis News & Views

Patient Newsletter    ⌘    Vol. 1, No. 4    ⌘    December, 2005



## Lyn Pickel Hired as Melorheostosis Association's First Executive Director



Lyn Pickel, who is a Founding Board Member and has served as Board President for the past 2 ½ years, has been hired by the Board to fill the position of Executive Director. In announcing Lyn's appointment, Alice Albin, interim board president, said:

"Lyn and the rest of the Board have felt for some time that the Association was nearing a crossroads. On the one hand, we are on the edge of finally possessing some definitive answers for melorheostosis. We have a world-class group of researchers and clinicians dedicated to helping find appropriate treatments and eventually a cure for this elusive disease. On the other, we have been consistently frustrated by our individual and collective lack of time and people resources to maintain the level of organizational momentum needed to achieve our mission. With Lyn's appointment as Executive Director, our organization will finally have the dedicated peo-

ple resources required to truly move our cause forward."

Lyn, a mother of an 8-year old daughter, Monica, with bilateral melorheostosis of the upper extremities, is a mother of 7 children, ranging in age from 22 to 8. She has an extensive background in administration, having served as a consultant in the area of organizational development for many not-for-profit organizations as well as a number of international corporations. She and her husband, Tony, live in St. Louis, Missouri.

Lyn will work for the first six months on a part-time basis and will report directly to the Board. In responding to the news of her appointment, Lyn said, "Being named Executive Director is the culmination of my dreams, efforts, yearnings and passion for the past seven years. At this point in our Association's evolution, our greatest effort must be directed towards creating a sound organizational/administrative infrastructure. We also must create a strategic plan to identify our needs as well as develop short- and long-term objectives. I am humbled by this magnificent opportunity, and I believe that together, with the membership, the Board, and our wonderful advisory panel, we will indeed achieve our mission."



## 2005 Season's Greetings



*The Association Board and a few of our friends from the 2005 Conference have the following wish for you:*

*May your holidays be filled  
with happiness,  
a loving family,  
wonderful friends, and  
countless memories!*

*Enjoy yourself and life's  
many blessings!*

### Association Mission...

*The Melorheostosis Association is a not-for-profit organization dedicated to finding the cause, treatments and cure of melorheostosis. Our focus will be on promoting greater awareness and understanding of this progressive disease and its manifestations through education, research, communication and advocacy efforts on behalf of those affected by it as well as those dedicated to alleviating it.*

## Board News

### ***Alice Albin Named Interim Board President***

Alice Albin, Association Secretary and a Founding Board Member, has been named to fill the vacancy of Board President, created when Lyn Pickel resigned recently to become Executive Director.



For the past 3 years, Alice has taken a lead role in coordinating a number of important organizational activities and projects. She did virtually all of the planning for the first international conference, held in East Lansing, Michigan, in 2004. Of her appointment, outgoing President, Lyn Pickel, commented: "Alice is a person driven by an uncompromising demand for excellence. She never does anything halfway. The conference that she put together in East Lansing set an exceedingly high standard of achievement which every other conference now strives to replicate."

Among her many other tasks, Alice is primarily responsible for the production of the newsletter, is bookkeeper and accountant, and is currently working on completing the organization's first brochure. She also is developing and compiling a book of organizational policies and procedures, a document which Lyn and the rest of the Board sees as vital in creating a strong organizational infrastructure for the foundation.

Alice, who has struggled her entire life with melorheostosis and two other related diseases—Osteopoikilosis and Osteopathia Striata—is the mother of five. She and her husband, John, live in Lansing, Michigan. Alice is employed as an Administrative Assistant at Michigan State University and has worked for the university for nearly 33 years.

Alice will serve the remainder of Lyn's term as interim President. Formal elections for the position of President will be held at the Board meeting in June 2006.

### ***Kathleen Harper Reappointed to Director Position***



At the July, 2005 Board of Directors Meeting, Mrs. Kathleen Harper's appointment as Director was unanimously extended for 3 years. Kathleen is also a Founding Board Member. She initiated and maintains the Association website. Kathleen's current term will expire in May, 2008.

Kathleen is busily involved in a major update of the website, and has been working closely with Dr. Pam Robey on establishing protocols for scientists to obtain samples from the DNA Tissue Repository.

## Scientific/Medical Advisory Panel News

### ***Our Heartiest CONGRATULATIONS on Scientific Achievement go to:***

**Dr. Michael A. Zasloff**, Professor, Departments of Surgery and Pediatrics; and Director, Surgical Immunology at Georgetown University, for his and his colleagues' discovery of a new class of antibiotics, found in fungi, proven effective in treating drug-resistant bacteria. Dr. Zasloff's discovery was published in the October 13, 2005 journal *Nature*. Those of you who attended the conference will undoubtedly remember that Dr. Zasloff treated us to an exciting sneak preview of his discovery and shared news of the impending announcement at Monday night's dinner.

**Dr. Michael P. Whyte**, Medical-Scientific Director for the Center for Metabolic Bone Disease and Molecular Research at Washington University, for his and his colleague's discovery of the cause of the extremely rare disease, x-linked recessive idiopathic hypoparathyroidism. Dr. Whyte's discovery was published in the September 2005 *Journal of Clinical Investigation*. It is believed that this discovery will lead to new treatments for the more common condition of osteoporosis.

### ***Surgical Management of Melorheostosis: General Information and Considerations***

#### ***Jeffrey C. King, M.D. and James Dobyns, M.D.***

Dr. King has written a document containing general guidelines aimed at educating patients and their doctors about issues related to surgical outcomes for melorheostosis patients. Dr. King based his findings on a retrospective review of 15 cases of surgery for the manifestations of melorheostosis found in the upper extremities, a review of the available English literature, a non-scientific review of 41 patient-reported results of surgery for melorheostosis, and his own professional experience.

The surgical document soon will be posted on our website ([www.melorheostosis.org](http://www.melorheostosis.org)). Patients contemplating surgery may find it useful to share this information with their doctors.

*Note: News and announcements regarding the Scientific/Medical Advisory panel members should be submitted to Lyn Pickel at [lynbpickel@earthlink.net](mailto:lynbpickel@earthlink.net) or Alice Albin at [albin@msu.edu](mailto:albin@msu.edu) for publication in the next newsletter.*

# Scientific/Medical Advisory Panel News (cont'd)

## List of Questions Submitted to Scientific/Medical Advisory Panel

Following the recommendation of Dr. Fred Kaplan, Chair of the Scientific/Medical Advisory Panel, the Melorheostosis Association membership recently compiled an extensive list of "most commonly asked questions" about melorheostosis. Dr. Kaplan now plans to distribute this list among the advisory panel members, assigning questions based on their content and panel members' respective areas of medical specialty and knowledge. Once all of the questions have been answered, the list will be posted as a helpful resource on the Association's website, <http://www.melorheostosis.com>. "The IFOPA Association did this," explained Dr. Kaplan, "and their membership found it to be an invaluable resource in providing a body of credible information on those questions and concerns that seem to surface most often."

While the list has now been sent to Dr. Kaplan, it's never too late to submit others! To view the current list of questions and/or to add others, visit the website and click on the link on the homepage entitled: "Have your Questions Answered by the Scientific/Medical Advisory Panel".

## Informational Brochure on Melorheostosis

Alice Albin, reports that the first informational brochure on melorheostosis is currently in the works, and following its review and approval by the Scientific/Medical Panel, will be placed on the website as a downloadable document. Alice said, "This brochure is intended to serve as an educational/informational resource on melorheostosis for patients and their loved ones, as well as for healthcare professionals who may have never encountered or need a fuller understanding of the disease."

The brochure provides an explanation as to what melorheostosis is, its rate of incidence, details information about diagnostic tools, describes some of the more prominent symptoms and complications, explains the difficulties in conjecturing long-term prognosis, details what is known to date about its cause, delineates treatment options and lists other related conditions.

Once the brochure is in final form, Lydia Zepeda, Treasurer of the Association, has arranged through her contacts at the University of Wisconsin to have it translated into several of the world's most commonly-spoken languages, making the Information easily accessible to our growing global audience.

*Be watching for notice of the brochure's debut on our website, <http://www.melorheostosis.org>!*

## Steady Progress Being Made Towards Establishing Repository

With the help of Dr. Pamela Robey, Chief, Craniofacial and Skeletal Diseases Branch of the National Institute of Health, the Melorheostosis Association is continuing its efforts to establish a central, neutral world-wide repository for melorheostosis samples.

In explaining the importance of establishing a neutral repository, Lyn Pickel, Executive Director, noted, "Being a rare disease makes melorheostosis exceedingly difficult to study. Researchers are limited not only by sparse funding, but also by the small size of samples available for study. The establishment of a cen-

tral repository at NIH would allow scientists from all over the world to apply for blood, plasma, and tissue samples contributed by a wide and diverse cross-section of melorheostosis patients.

Distribution of samples will be overseen by a newly-appointed scientific review panel to ensure that samples are used for legitimate and relevant research purposes. So far, Dr. Robey and Dr. Drew Mannes, Staff Clinician, Pain and Palliative Care Service of NIH, have agreed to serve on the review panel. The Association is currently soliciting a third and final member for this committee.

This past summer, participants attending the Melorheostosis Conference eagerly donated their plasma in order to provide an initial "deposit" for the NIH repository. Dr. Michael Whyte and Dr. Geert Mortier, members of the Scientific/Advisory panel, have already applied for these samples in order to conduct further mutational analysis of melorheostosis in an effort to discern its cause.

If you are interested in donating blood or tissue to NIH, please contact Lyn Pickel ([lynbpickel@earthlink.net](mailto:lynbpickel@earthlink.net)), Alice Albin ([albin@msu.edu](mailto:albin@msu.edu)) or Kathleen Harper ([Kathleen@harpervision.com](mailto:Kathleen@harpervision.com)) for specific, step-by-step instructions. NIH will have you sign a consent form (if a child is involved, two consent forms must be signed), and furthermore will need your doctor's contact information and cooperation in order to provide instructions for preparation and shipping.

Please understand that donating samples requires some time and work on your part in obtaining your doctor's cooperation and approval. As such, please inform us right away of your anticipated surgical date if you plan to donate your blood or tissues.

## Become an Advocate for Bone Research

Dr. Laura McCabe, Associate Professor in the Department of Physiology at Michigan State University and member of the Scientific/Medical Advisory Panel, recently helped launch a website explaining how to become an effective advocate for bone research. The site, <http://www.asbmr.org/about/committeefinfo.cfm#Women's%20Committee>, was established to promote awareness and garner support for increasing bone research funding. The site contains an informative and a downloadable brochure describing what you can do to promote bone sciences. It also has power point slides detailing the essential role of advocacy in securing musculoskeletal research funding.

The site contains a link to the website for the American Societies for Experimental Biology (<http://www.faseb.org>) where it is possible to simply punch in your zip code in order to forward a form letter to your congressional representative supporting bone research. According to Laura, "Many researchers are linked to this site and send letters to their congressional representatives on a regular basis to show support for biomedical research. I think the more people who participate, the better!"

The progress of bone research depends on all of our continued and focused support. Scientists and patients make the best advocates for biomedical research. So, check out the website and become a vocal advocate for bone research by sending a letter to your congressional representative today!



# Scientific/Medical Advisory Panel News (cont'd)

## 5 Facts Concerning the Future of Bone Science Research

1. US National Institute of Health (NIH) funding cut-offs are dropping.
2. More specifically, bone research is under-funded in relation to the economic burden of bone-related diseases.
3. Scientific process and progress—including peer review and animal research—are under threat from vocal minorities.
4. Disease-focused advocates compete for funding their causes.
5. Scientists and patients make the best advocates for biomedical research.

-excerpted from the American Society of Bone and Mineral Research website

## News from Our Members

**Note:** Beginning with this edition of *News and Views*, we are including a column devoted to news about our members. If you would like to share recent news about yourself or your family, please submit information to either Lyn Pickel ([lynbpickel@earthlink.net](mailto:lynbpickel@earthlink.net)) or Alice Albin ([albin@msu.edu](mailto:albin@msu.edu)). We welcome photographs to accompany your notes!



Mari Robinson, now Mari Robinson Painter, was married to Tom Painter on August 18, 2005, in Bismark, North Dakota. Mari, her son, Matt, and Tom, attended the melorheostosis conference this past summer. Of Tom's attendance at that conference, Mari observes: "Tom found the conference to be very informative... and a way for him to better understand the disease and its effects. It was a way for him to understand why I have the limitations and pain that I do. It is tough for someone that cares for someone else to comprehend the disease without any education. Because of it, I think it was just as important for patients as it was for spouses and family members."

## 2006 Conference Planning & News

### 4th International Melorheostosis Association Conference

June 4-6, 2006

University of Wisconsin—Madison

Conference Host: Lydia Zepeda, Director

Hotel: Best Western Inntower—Madison

Phone: 1-608-233-8778

Fax: 1-608-233-1325

Email: [info@inntower.com](mailto:info@inntower.com)

Website: <http://www.inntower.com>

Group Rate: "MELO"

\$62 Single

\$82 Double

*In the wake of 2 extremely successful and rewarding conferences, the 2006 conference promises to be even bigger and better! Planning is underway to schedule speakers, and to facilitate another invigorating exchange between both medical/scientific professionals and patients alike.*

Formatting changes are being considered in order to ensure patients continue to learn and benefit from their participation. If you have any suggestions for topics or activities, please contact Lydia ([lyzepeda@uwisc.edu](mailto:lyzepeda@uwisc.edu)), Lyn or Alice. We are striving to cover some new topic areas, and welcome any input that you may have.



Registration documents will be available soon. Feel free to make your hotel reservations now!

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About the 2005 Conference in  
St. Louis, MO

**Abstracts & Slide Shows** – Abstracts from the scientific talks are available on the website. Wherever possible, a hotlink is available to show the speaker's slide presentation. Most are heavily scientific (and not for the faint of heart), but are very much worth viewing!

**Event Photos** — See page 7 of this newsletter.

# The Fundraising Page

## HELP US HELP YOU!!!

One of the biggest challenges the Board has faced is in trying to inspire patients and family members to fundraise. We need money to operate, fund the conferences, and to pursue our mission. The conferences have been extremely beneficial to our cause, and have established a fast-paced momentum with our Scientific/Medical Advisory Panel. Lack of funds will jeopardize this momentum. Members of our Panel are asking for financial support to conduct research on melorheostosis. **We need your help in building our cash reserve. Please do a fundraiser. Your fundraiser will make a difference! We need everyone's involvement—please don't rely on the 'other guy' to take care of this. We are all in this together!**

The Association is licensed to solicit funds in the following states:

California	New York
Florida	Oklahoma
Illinois	Pennsylvania
Massachusetts	Washington
Michigan	Wisconsin
Missouri	

If you want to fundraise in a state other than those listed, please contact Alice Albin so an application for license can be made.

Gift envelopes can be made available for your use by contacting Lyn or Alice. If you choose to prepare your own envelopes, please make sure donors forward their contributions to the Association Office and write your name on the backside of the envelope so we know which fundraiser to credit. This way we can let you know who has made a contribution. Again, we will help in any way we can. Many, many thanks in advance!!!

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### A Fundraising Example

This is the 2005 holiday letter Lyn Pickel prepared and sent out to friends and members of her family. This could be easily modified and used by any member of this association.

Dear Friends,

It has been another remarkable year for the Melorheostosis Association. With the help and generosity of our many friends and donors, we convened a world-class conference—international in scope—at St. Louis University, this past July 17-19<sup>th</sup>. The meeting featured an impressive array of researchers and clinicians from some of the most prestigious universities and medical facilities in Belgium, England and the US. It also attracted a wonderful cross-section of patients—children and adults—who came from as far away as Europe and Canada.

The discussion that occurred between the scientists, clinicians and patients who attended that meeting resulted in

a virtual explosion of ideas and led to a number of exciting collaborative research initiatives involving advanced mutational analysis, animal model studies and a long-term historical review of all melorheostosis cases, now being conducted by the head of research for Shriner's Hospital world-wide in Montreal.

The conference also afforded a rare opportunity to collect blood/DNA samples from our patient population. These samples were then sent to the National Institute of Health where they will be stored and eventually distributed to scientists around the world for research purposes.

In just a few weeks, we will have our first official informational brochure about melorheostosis posted on our website, [www.melorheostosis.org](http://www.melorheostosis.org). The brochure will be formatted as a downloadable document so that it can be easily accessed and distributed to patients and doctors seeking the most current and reliable facts about the disease. In addition, we are in the process of compiling a book of the most commonly-asked questions about melorheostosis. The book, to be published next year, will contain answers contributed by members of our scientific/medical advisory panel who collectively represent the foremost researchers and clinicians in the field of bone study and treatment.

Our next conference, to be held at the University of Wisconsin at Madison, June 4-6, 2006, promises to be just as stimulating and productive as the last two. Currently, we are in the beginning stages of planning the event. Our esteemed scientific/medical advisory panel has eagerly accepted our invitation to speak again, and we are busy lining up topics and some new speakers who will appeal to the needs and concerns of our growing patient population.

Of course, it goes without saying that all of these initiatives require a substantial amount of money. We have been so blessed by the generosity of our donors over the past three years. Their contributions have made our phenomenal progress to date possible. But continued and increased funding will be needed not only to sustain our organization and related patient needs, but to hopefully provide seed money for the research that will be required to ultimately conquer this insidious, disfiguring and painful disease.

This holiday season, we ask you to please consider a gift to help the Melorheostosis Association continue its good work and progress. You can be assured that every dollar donated benefits the children and families affected by this orphan disease and provides momentum for finding definitive answers for its treatment and eventual cure.

Blessings to You and Yours during this Holiday Season!

Lyn Pickel



Stacey,  
Terry  
&  
Harry

## Getting to Know You

### The Cole Family

Harry, Theresa, Suzanne & Stacey

**Note:** This is the first in a series of articles introducing one of our members or families to our growing melorheostosis community.

**Tell us a little about your family. Terry:** I am 53; my husband, Harry, is 54. We have two daughters, Suzanne, 19, who is away at college in her sophomore year; and Stacey, 17, who is in her junior year of high school. We live in Milltown, New Jersey. Harry and I just celebrated our 25<sup>th</sup> wedding anniversary this past October 11<sup>th</sup>.

**What do you and Harry do for a living, and how long have you been doing this? Terry:** I am a Commercial Property/Casualty Insurance Account Executive. I have been in the industry for 33 years; at my present job in Princeton for 19 years. Harry has been in the freight forwarding business for 35 years, starting out at Federal Express in the '70's when it had its humble small beginnings. He is currently working for a small company in Carlstadt, New Jersey, specializing in unique freight forwarding situations.

**How far along is Stacey in high school? What is her favorite subject? Her favorite activities? What plans does she have following high school? Does she have any idea as to her eventual career goals? Terry:** Stacey is an Honor Student in her Junior Year of High School at Spotswood High School, NJ. Her favorite subjects are Creative Writing/Journalism and Art. Her favorite activities are reading, building her own DVD library, poker tournaments, and drawing. She is facing 2 more surgeries, one this coming June and another after senior year. She is considering delaying college for one semester so she would be all through the surgeries, but has not yet decided completely on this. Her main priority is to get through junior year, visiting colleges, and taking her SAT's. She thinks she would like to pursue Creative Writing/Journalism.

**What sort of activities/hobbies does your family enjoy doing together? Terry:** We enjoy eating out once or twice a month, going to the movies, and playing cards.

**What is your connection to melorheostosis, and how did you hear about the Melorheostosis Association? Terry:** Stacey has a combination of Bushke-Ollendorf Syndrome (a combination of osteopoikilosis and connective tissue nevi) and melorheostosis. During the

process of working towards Stacey's medical diagnosis in her childhood, Harry discovered he had Bushke-Ollendorf syndrome as well. I heard about the Melorheostosis Association while looking for any current information on the internet.

**What have been the greatest challenges you have confronted, individually and as a family, relative to melorheostosis and these related conditions?**

**Terry:** The greatest challenge we have confronted was the uniqueness of Stacey's disorder, in that she had two such rare bone disorders. Stacey was already being tested at age 3 because of the severe joint contractures forming in her right foot, ankle and knee. Doctors were perplexed, and what were the odds of having one rare disorder, let alone two? At the time Stacey was diagnosed first with the BOS, we were told there were only 72 medically known cases in the world. All this before the introduction of the internet! At the time, I had a cousin working at Johns Hopkins who would research the medical archives there for anything and everything he could find for us. So the first greatest challenge was just finding any kind of information about this, and going up and down emotional roller coasters because the doctors were learning right along with us. The other biggest challenge for us as a family was to care for Stacey through her surgeries, while still working full time and juggling all our other responsibilities. Fortunately, I work for a wonderful firm that allowed me flex hours during these times which was a huge help. Stacey's biggest frustration was enduring so many surgeries, only to have some of the same problems re-occur. The process was long and difficult for her. She was in an Ilizarov Fixator for 10 months and would endure rigorous, painful physical therapy seven days a week.

**What is your greatest hope relative to the work of this Association? What sort of services/activities would you like to see the Melorheostosis Association offer?**

**Terry:** It is our hope that the Melorheostosis Association can provide a clear, concise, user-friendly central database to offer assistance for people, as well as establish a support group of people that can easily communicate with each other. It has already been a godsend to us, and we feel blessed to have been a part of the conference held in St. Louis this past July and to finally meet others who can share their experiences. Knowledge is power, and the more we help each other learn about this mysterious disorder, the more empowered we can be.

**Any words of wisdom or inspiration that you would like to share with our membership given your experience with this disease? Terry:** Try to remain focused on what makes you happy, and pursue your dreams. Don't let the frustration that we all encounter regarding this take over your life.

**In our next issue...  
An Interview with...  
"Kevin Amelio Collevecchio"**



A publication of the Melorheostosis Association  
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*A few photos from the  
3rd International Melorheostosis Association Conference, St. Louis, MO*



Dr. Deborah Wenkert



Linda Hembree



Mrs. Shaughnessy & Alice Albin



Harriet & Robert Bradney (Lyn's parents)  
Lyn & Tony Pickel



John & Dawn Davidson



Clarice & Sarah Stokes



Kathy Jerome & Carolyn Parker



Kathleen Harper & Dr. Jeff King



Dr. Michael Zasloff &  
Dr. Fred Kaplan



Dr. Laura McCabe & Kathy Ryan



Dr. Geert Mortier



Dr. Roger Smith



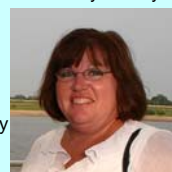
Stephanie Papke, Dan Papke, Andrew Carnell, Mykel Papke, Paul Sowerby



Madelyn Reilly & Debbie Tharpe



Rob & Scott



Donna DeLuca



Ann & Faye



Michelle Lundie



Odette Blanchard, Frankie Mesa, Alice Albin



Dr. Michael Whyte & Dr. Steven Mumm



Lyn Pickel & Dr. Mitch Rottman



Martine Phipps & Tara,  
Amelio Collevchio



Monica, Clarice & Maureen



Lydia Zepeda & Mishka



Mr. & Mrs. Joseph Shaughnessy and Dr. & Mrs. Rob Fleming



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**"Nature is nowhere accustomed more openly to display her secret mysteries than in cases where she shows traces of her workings apart from the beaten path; nor is there any better way to advance the proper practice of medicine then to give our minds to the discovery of the unusual law of nature, by the careful investigation of cases of rarer forms of disease."**

**William Harvey, 1657**