

Melorheostosis News & Views

Patient Newsletter ⌘ Vol. 1, No. 1 ⌘ November, 2004



www.melorheostosis.com

The President's Corner...

It is with great pleasure that we bring to you the first newsletter published by the Association. Our plan is to publish this quarterly so we can keep you informed of the many activities underway on behalf of melorheostosis patients worldwide. I hope you find this newsletter useful, and you will share how we can improve upon getting information back to those who really need it—the patients!

As you may have guessed, this past year has been a whirlwind—particularly for Kathleen, Alice, Linda and me. Silly us: we thought once we got past the conference, it would be smooth sailing. Oh, to the contrary, my friends. One of the main realizations the 4 of us took away from the conference was how much work there is to do in maintaining our momentum with the scientific community, and in simply defining and organizing the tasks to be undertaken by the Association. As you read through this newsletter, you will learn of the many projects we are working on.

This Association is definitely on the fast track in terms of sparking interest in our cause. Not only do we have the best scientific leader in Dr. Fred Kaplan, but we have a stellar and interested group of people on our Panel who are excited about studying melorheostosis, and genuinely interested in helping us. This, coupled with the fact that the faulty gene involved in creating melorheostosis has been identified, is causing all kinds of additional enthusiasm for our cause. I am pleased to report that Dr. Geert Mortier, who is on the team that discovered the melorheostosis faulty gene, has agreed to speak at our next conference, and has enthusiastically accepted our invitation to join the Scientific/Medical Advisory Panel. Another scientist in Berlin, Dr. Berthold Struk, is conducting genetic studies on melorheostosis and osteopoikilosis. We are hoping he will also agree to speak at our next conference. **And**, one of our very own Scientific/Medical Panel members, Dr. Michael Whyte, is laying the groundwork to begin a research study on melorheostosis. Our Association is blessed to be in the middle of these exciting breakthroughs, and also on the verge of witnessing many more projects as a result of Dr. Mortier's breakthrough discovery. I, along with many other Board Members, believe the results gained from the study of melorheostosis will lead the way to discoveries relevant to many other disabling musculoskeletal bone diseases. I am thrilled to be a part of all this and I hope you are too!

I want to take an opportunity to express how desperately we need people to conduct fundraising campaigns. There will be more about fundraising later in this newsletter, but I do want to personally stress how everything our group 'does' or 'tries to do' boils down to 'how much money we have.' **Please, I urge everyone to conduct at least one fundraiser.** Alice, Donna and I will give you ideas and/or provide you with whatever information you need to conduct a successful campaign. We genuinely need your help!

Again, I hope you enjoy this newsletter. Please feel free to share whatever comments you feel will help move our Association along. We are counting on your input and participation. Thanking you in advance, *Lyn Pickel*.

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An Exciting Recent Development...

Thanks to the magnanimous efforts of Andrew Carnell, Paul Sowerby, Martine Phipps, and Michelle Lundie, a charitable U.K. Melorheostosis Association is on its way to being established. To date, charity status has been applied for and fundraising has begun. The U.S. and U.K. Melorheostosis Associations will work together in joint pursuit of finding the cause, treatments and cure of melorheostosis. Our hats are off to the U.K. team! Thank you one and all!

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.

About the Board of Directors...

Lyn B. Pickel, President and Founder, is a wife and mother of 7 children (4 at home; 3 in college) and resides in St. Louis. Lyn is a social worker and private consultant to industry.

Alice J. Albin, Secretary and Founder, is a wife and mother of 5 children (3 at home; 2 teenagers and 1 finishing law school). Alice is employed full-time by Michigan State University.

Kathleen D. Harper, Director, Webmaster and Founder, is a wife, has a son in college, and resides in New York. Kathleen is a lawyer by trade but now devotes a significant amount of her time to charitable endeavors, including—lucky for us—melorheostosis!

Andrew Carnell, Director, is a husband and father of 3 young children, and a private entrepreneur. Andrew's U.K. businesses relate to web marketing and real estate development.

Donna DeLuca, Director, is a wife and mother of 4 (ranging in age from teenager to baby) and resides in the Boston area. Donna is a day care provider and is very active with youth groups.

Lydia Zepeda, Director, is a wife, mother of 2, and a Professor of Economics at the University of Wisconsin. Lydia has accepted the open Treasurer position.

Michelle Lundie, Director, is a mother of 3 young children and resides in the U.K. She is employed in the health care industry, and brings with her an eagerness unsurpassed to educate doctors about melorheostosis.

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Announcing 2 Open Director Positions

The Board is accepting nominations for 2 open Director Positions. If you are interested in serving the Association in a formal capacity, please contact one of the current Board Members for more information.

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Regretfully, Linda Hembree, who is one of the original 4 Association Founders, resigned her post as Director in September. The Board wishes to collectively acknowledge Linda's dedication and contributions during our first organizational year, and express a sincere **thank you** for everything she has done to help us get started. Linda is still very committed to our cause and promises to help whenever the need arises. **Thanks again, Linda!!**

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Personal Reflections on the 2nd Annual Conference of the Melorheostosis Association
“Symposium Presentations and Selected Patient Case Studies with the Scientific/Medical Advisory Panel”

August 24-26, 2004

by Lyn Pickel

Michigan State University

So many of you have inquired about how the 2nd Annual Melorheostosis Conference went. I have personally answered this question by describing it in such broad terms as: “moving”, “unbelievable” and “awesome”. It was indeed all of these, but such words leave a lot to the imagination. Therefore, I am taking this opportunity to fill in some of the details for those of you who were not able to attend this year.

The Conference was scheduled to begin on Tuesday, August 24th, but due to the tremendous anticipation shared by participants and our Scientific/Medical Panel alike, many of us arrived a day early. The joy of meeting one another after months—and in some cases, years—of trading messages back-and-forth over the website bulletin board was... well, indescribable. My mother, who so generously accompanied me in order to take care of Monica, likened the experience to what must transpire when an adopted child finally meets his/her biological mother or father. There was an instant bonding and recognition that could only take place among those who have shared exceptional life circumstances.

Several of us spent the day prior to the conference’s start sharing our stories with one another and showing and discussing our x-rays and medical histories with Dr. Fred Kaplan, Chair of the Scientific/Medical Advisory Committee. The conference officially began on the evening of the 24th with a lovely wine reception. Although the emphasis of the event was definitely social, the entire group ended up sitting in a large circle with Drs. Kaplan, Shore and Whyte, discussing melorheostosis: its seemingly random occurrence, its vast difference in presentation from one individual to the next, its potential link to osteopoikilosis, whether it may be hereditary, and on and on. One could almost see the wheels in Drs. Kaplan’s, Shore’s and Whyte’s heads turn as we talked late into the night. Carolyn Parker, a frequent participant in web site discussions, shared the exciting news that her family and she are participating in a study being conducted by Dr. Struk of Berlin. The study, in part, is looking at the connection between melorheostosis and osteopoikilosis. Much to our shock, Carolyn revealed that 70 members of her extended family have osteopoikilosis and one of her first cousins also has melorheostosis. The Scientific/Medical Panel was enthused about the implications of such a study, noting that the large size of the family and prevalence of osteopoikilosis will facilitate identification and future study of the gene(s) responsible for these diseases.

During the following day, participants were privileged to have several members of our Scientific/Medical Advisory Panel present reports. Dr. Michael Whyte, Medical-Scientific Director, Center for Metabolic Bone Disease & Molecular Research at Washington University, gave a fascinating talk on the links between melorheostosis and a number of other sclerosing bone conditions. Some of these diseases, such as Pagets, are better understood and more widely researched than melorheostosis. As such, they potentially hold important clues for better understanding the disease process involved in melorheostosis.

Dr. Jill Helms, Associate Professor, Department of Plastic and Reconstructive Surgery at Stanford University, gave a presentation on the etiology of melorheostosis: clues from developmental biology. One of the curiosities of melorheostosis is that it tends to predominantly affect the limbs but rarely is found in the skull. The fact that the embryonic skull is formed from a different type of cell than those that create the developing limb may explain why this is so. But why one type of cell is more susceptible to this disease than another is unknown. Dr. Helm’s research is directed at resolving this question.

Dr. Pam Robey, formerly of our panel and Chief of the Craniofacial and Skeletal Diseases Branch of NIH, discussed stem cell biology in reference to understanding the origins of melorheostosis. She taught us about stem cells, how they work, and the importance of stem cells in research. Dr. Robey fielded some amazing questions from patients and Panel members alike. Dr. Robey emphasized the importance of creating and maintaining a patient registry for researching and gauging the demographics of melorheostosis. [Since the conference, Dr. Robey has indicated her willingness to guide the Association in establishing a patient registry. We plan to discuss this issue further with her in the upcoming weeks. At the same time, we plan to explore the possibility of setting up a central, neutral repository where patients could submit their DNA, blood, and tissues for research purposes.]

Dr. Eileen Shore, Research Associate Professor of Orthopaedic Surgery at the University of Pennsylvania, talked about her and her colleagues’ work on both FOP and POH. These particular bone diseases are destructive and devastating beyond words, and ultimately result in premature death. Her group continues to pursue the hunt for the gene responsible for FOP (this disease causes soft tissue to turn into bone). Her team’s relentless work has yielded some intriguing possibilities as to why bone metabolism can sometimes go terribly awry, but as of yet, the culprit gene(s) for FOP has eluded detection.

Dr. Jeff King, the newest addition to our Scientific/Medical Panel and Clinical Associate Professor at Michigan State University, gave an informative presentation on the orthopaedic aspects of melorheostosis. Dr. King has analyzed results on the surgical outcomes of numerous melorheostosis patients. His statistics and findings, which underscore surgery’s general lack of effectiveness in addressing melorheostotic problems, indicate a pervasive lack of knowledge on most doctors’ parts regarding this disease and how best to treat it. Several of the participants recounted their own unsuccessful experiences with surgery. Dr. King requested that these individuals secure their surgical notes for him so that he can add them to his study. He hopes to publish his study in the near future, providing clinicians and surgeons with a more definitive guide as to the circumstances under which surgery is most likely to prove successful in mitigating symptoms.

At dinner on the evening of the 25th, Dr. Fred Kaplan, head of our Scientific/Medical Panel and Chief of the Metabolic Bone Diseases and Molecular Medicine at University of Pennsylvania, gave a stirring keynote speech on the search for answers to melorheostosis. Dr. Kaplan recounted how, as a college student, he had managed to sneak into an abandoned hospital which had been built to house Tuberculosis patients. This hospital was filled with row after row of “brand new” iron lungs, never used. His poignant message was not lost on the group: a cure for TB had been found before the construction on the hospital was finished. Dr. Kaplan’s story was inspiring and hopeful. The search for the answers to melorheostosis, albeit fraught with frustration, will culminate in a similar ending someday, with the discovery of a cure or effective treatments. At that point, our Association will happily disband.

On Thursday, the 26th, several members of the Scientific/Medical Panel met with each participant individually to look at their x-rays and MRI’s, to hear their stories related to melorheostosis and to ask questions about different aspects of their treatment and medical histories. These sessions were not intended to be treatment-oriented, but rather represented an opportunity for the researchers and doctors to glean observations, ask questions and to generally piece together a

better understanding of this puzzling disease. As Dr. Kaplan remarked: “It is rare that a doctor—even in the specialized area of orthopaedics—would stumble across more than 1-2 cases of melorheostosis in his/her professional lifetime. The number of patients we have seen today represents a veritable wealth of new and revealing information!”

While Drs. Kaplan, Shore and Helms were busy talking to patients, faculty members from MSU gave informative presentations on a variety of subjects. The following briefly summarizes the wealth of knowledge these individuals shared with patients.

Dr. Laura McCabe, Associate Professor of Physiology and Radiology, spoke on the response of bone dynamics to exercise, aging and disease. Dr. McCabe taught patients about the normal bone remodeling process and compared the normal process to what happens with other bone disorders such as osteoporosis. This talk provided patients with a visual perspective on how bones grow and stimulated discussion on what could be going wrong with melorheostotic bone.

Dr. Mark DeLano, Associate Professor of Radiology, educated us on radiological imaging, giving constructive advice on the amount and type of radiation that is dispensed as one is subjected to various imaging tests. While Dr. DeLano stressed that all forms of current testing are safe (x-rays, MRI’s, bone and cat scans), he encouraged patients to refrain from having tests *if* it is unlikely that the imaging test will result in an amended treatment plan. In other words, he encouraged patients *not* to get x-rays every 6 months just for the sake of getting x-rays.

Dr. Jeffrey Kovan, Director of MSU Sports Medicine, spoke on the benefits of physical therapy in establishing improved function and pain reduction. Dr. Kovan said that the ‘no pain, no gain’ philosophy has been abandoned, and strongly encouraged patients *not* to ‘work through pain’. He said, ‘pain means—stop’. Dr. Kovan encouraged stretching exercises to increase range of motion. For pain management, he suggested use of an electrical nerve stimulation device (TENS Unit), medical patches (Licoderm, thermal wraps), and topical anesthetics.

Dr. Rachel Fisher, Professor of Pediatrics and Human Development, spoke on how genetics can help us understand disease. Dr. Fisher defined in layman’s terms the words commonly used by geneticists like proteins, DNA, genetic code, mutations and chromosomes. She then explained how genes are passed along to offspring by using a ‘fat cat’ model (you had to have been there!). She also explained how a genetic disease can be both inherited and sporadic. Dr. Fisher’s talk shed new light for everyone in relation to the genetic-based talks which were heard the day before by Drs. Whyte, Helms, Shore, Robey, Kaplan and King.

Dr. Janet Osuch, Professor of Surgery, gave a touching account of her personal battles with cancer, her many years experience in advocacy, her involvement with breast cancer initiatives, and a brief overview of the role epidemiology (the number of estimated incidences of a disease) plays in securing resources. Dr. Osuch stressed the importance of developing policies and procedures in order to maintain an effective advocacy organization. She cautioned our group about many organizational pitfalls to avoid (such as bad press coverage due to financial mismanagement), and about how groups who do not take the time to properly document and think out the governing rules of their organization generally fail.

Each day, we also had the benefit of having a keynote speaker at lunch. On Wednesday, Dr. Cam Riessinger, Psychologist, Sparrow Health Systems, spoke on the topic of pain management (an issue of pressing importance to most of our participants). She explained how chronic

pain can be a huge problem for not only the patient, but the family as well. Dr. Riessinger gave many helpful tips for coping with pain, and strongly encouraged patients to seek help from local pain clinics. Most clinics take a multidisciplinary approach to treating patients suffering from chronic pain, including physicians who specialize in pain management, nurses, psychologists, physical therapists, etc.

On Thursday, Dr. Elizabeth Alexander, MSU University Physician, spoke eloquently on the dilemma of finding the “right” doctor when one is confronted with a rare disease and how best to establish a productive patient-doctor relationship. Her topic was “Unusual Diseases, Ordinary People, and Finding the Right Fit.” Dr. Alexander cleverly reminded us that doctors are people too, and as such, can sometimes be as overwhelmed as the patient when faced with dealing with a rare and mostly undefined disease. Dr. Alexander encouraged patients to request a ‘double appointment time’ if there were many topics to discuss with the physician, rather than trying to fit a long list of topics/ailments into a 15-minute slot. Interestingly enough, this talk touched not only the patients, but the Panel doctors as well.

The conference formally ended late Thursday afternoon, but several participants extended their stay until Friday, the 27th, not wanting the week and the special camaraderie to end. When the good-byes were finally said, there were hugs and tears all around. The comfort in knowing another conference is planned for next year helped as the participants and doctors bade their farewells.

The Conference—by all measurements—was successful beyond our wildest dreams. It brought so many of us together: names on the melorheostosis website bulletin board before, lifetime friends now. It assembled a world-renowned group of scientists, facilitated their thinking, challenged their assumptions, created new impressions and generally provided a rich source of information for proceeding with more enlightened study.

None of this would have happened were it not for the able and tireless efforts of Alice Albin, Board Secretary. Alice worked non-stop from April on, setting up the conference facilities, identifying and coordinating the presenters, planning the meals, arranging the Advisory Panel’s travel, constantly trouble-shooting and serving as our fearless task master. Words cannot begin to adequately express our appreciation for all her efforts. Alice also received tremendous help from Dr. Laura McCabe, MSU Associate Professor, who helped in planning and arranging the MSU speakers. The Association is indebted to her as well.

As I recall this historic conference, further research efforts are unfolding at breathtaking speed. In Belgium, Dr. Geert Mortier, along with a group of 20 other scientists, have discovered the gene responsible for causing melorheostosis and osteopoikilosis. Dr. Mortier has expressed his willingness to meet with our group to present his data. He has also enthusiastically accepted our invitation to join the Scientific/Medical Advisory Panel. During this next year, he plans to perform further mutational analysis of this gene in additional patients. Dr. Pam Robey is directing our efforts to establish a worldwide bone, blood and tissue repository. And in Berlin, Dr. Struk’s important work on understanding osteopoikilosis and melorheostosis continues, with the potential for shedding more light on melorheostosis’ elusive nature.

It was a wonderful and historic conference, and next year’s will be even better, given the promising strides in research that have been made recently. *We hope that everyone will be able to attend the 3rd Annual Melorheostosis Conference, time and place to be announced once Dr. Mortier informs us of his availability.*

Other Announcements and Developments...

Kathleen Harper has met with representatives from the National Organization of Rare Diseases (NORD). The Melorheostosis Association has applied for Associate Membership in this organization, and soon there will be a fact sheet posted on the NORD website about melorheostosis (www.rarediseases.org). Membership will entitle our Association to attend educational conferences on managing non-profit organizations, rare diseases, and securing funding.

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Dr. Jeffrey King is working on writing a summary of 'surgical considerations' for melorheostosis patients to use as a reference when contemplating surgery. This document will be posted on the website.

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Lyn Pickel and Dr. Pam Robey are diligently ironing out the details of establishing a central, neutral repository for banking DNA, blood and tissue samples. The rationale of a central, neutral repository is that it allows all researchers equal opportunity to apply for samples, thus spurring diverse research. A subset of the Scientific/Medical Advisory Panel will be asked to review requests for samples. NIH has informed us that the cost to house and manage sample dissemination runs about \$20,000/year.

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Donna DeLuca and Alice Albin have been working on an informational brochure and gift envelope. The envelope is ready for printing.

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Alice Albin has agreed to establish the accounting and gift acknowledgments on QuickBooks. Alice prepared the paperwork to several states in order to legally conduct fundraising. And, Alice prepared a preliminary draft of the Association Board Policies and Procedures Manual.

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Association Committee Assignments

Executive Committee – Lyn Pickel, Alice Albin

Fundraising Committee – Alice Albin, Donna DeLuca (Co-Chairs)

Communications & Awareness Committee – Kathleen (Chair), Donna DeLuca, Paul Sowerby (Paul is a colleague of Andrew Carnell)

Conference Committee – Alice Albin

Scientific/Medical Research Initiatives

Committee – Lyn Pickel, Kathleen Harper (Co-Chairs)



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How you can help ...

The long and short of it is we need all kinds of help. We need people to help us further organize our group, prepare publications like this newsletter, fill the open Director positions, contribute information to the website, etc. These tasks take time and could be better accomplished if we were aware of some of the skill sets of our fellow melorheostosis patients and able-bodied family members. *Would you be willing to help us pursue our mission by getting involved in the organization?* If so, please contact any of the current Directors.

Fundraising...

Food for thought... It costs a lot of money to run a non-profit agency. Even though a good percentage of the work can be accomplished through volunteer efforts of the Board, Panel, and others, this is *not* the most effective way to run a non-profit organization. In fact, Dr. Janet Osuch warned us vehemently that the original organizers of this agency will get burnt out if funding is not secured to fund regular positions. Also, she said that by law non-profits usually can only dedicate about 2-9% of their fundraising revenue to paid staff. This should give you an idea of how many dollars we will need in the bank before we can hire 1 or 2 people.

More food... Funding research will be very, very costly. Now that the melorheostosis gene has been identified, researchers are already asking if we have the resources to fund projects. Funding one postdoctoral level scientist at a lead university would cost approximately \$75-100,000/year!

And more food... As indicated earlier, we expect our tissue/DNA sample repository fees to be on the magnitude of \$20,000/year. It is possible that we can get a grant for this, but there are no guarantees when applying for grants, so ultimately we need to start building a cash reserve. Also, it is anticipated the 2005 conference will cost on the order of \$15,000.

In summary... In order to run a *top notch charity*, we must garner funding to hire an Executive Director and an office assistant. Keeping the charity in compliance with IRS regulations, state regulations, and the multitude of details involved in managing a non-profit are unbelievable. Simply put, it is very difficult to accomplish everything that needs to be done in a timely basis when 100% of the work is left to volunteers who already have families and full-time jobs. Over the short run, this will work—but over the long haul, it will not.

Where we are today... Thanks to Lyn Pickel and Donna DeLuca who raised about \$14,000 last summer, we were able to offset fully the costs of our 2004 conference, state registrations, and accounting software. We have about \$5,000 left.

Please get involved in fundraising. There are any number of opportunities from small scale to large scale (golf outings, bake sales, garage sales, auctions, raffles, personal letters to friends and family, youth group activities, dinners, etc.). If you happen to know a celebrity, this could be very helpful. You will be amazed at how generous people are when you reach out with a worthy cause at hand! Please contact Alice Albin or Donna DeLuca if you are interested in obtaining information and help on conducting a fundraiser. With the holidays rapidly approaching (and tax year-end), this is an excellent time to conduct a personal campaign!

In the meantime, Lyn and Alice are writing a grant to the NIH to try and secure funding for the 3rd Annual Conference. Alice (with the help of Lyn) will conduct a corporate fundraising campaign to pharmaceutical companies. Donna is working on a possible 'bracelet' campaign. Lyn and Alice also plan to conduct personal fundraising campaigns to family and friends this holiday season.

“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 than 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