

Melorheostosis News & Views

Patient Newsletter ⌘ Vol. 1, No. 2 ⌘ February, 2005



www.melorheostosis.com

The President's Corner...

Have you ever looked at our logo and wondered about its significance?

The logo depicts a distant sun spreading its radiant light through an otherwise dark void. The symbol is meant to convey the awesome power of hope in illuminating the darkest corners of isolation and despair. Anchoring this emblem is a triangle bordered by three words that capture our collective and deepest hope: "cause", "treatment" and "cure".

As I write this article, I am humbled by the miraculous events that have transpired over the last few months—events that have brought renewed meaning to this powerful imagery. We are blessed with four additions to our Scientific/Medical Advisory Panel. They are: Dr. Geert Mortier, Dr. Howard Worman, Dr. Peter Klein and Dr. Laura McCabe. A commentary on each of these spectacular scientists is included on page 4 of this newsletter.

Plans for the conference are shaping up very quickly. This conference will be interesting, and has all the makings to prove historic as well. With all these brilliant minds gathered in one place, at one time, just imagine the synergy for creating enlightened scientific discussion and sparking new insights! We are, my friends, on the cusp of great discovery. I truly believe that this occasion may provide the catalyst for illuminating the answers that have for so long eluded this disease. You are not going to want to miss this meeting!

This year we are planning a few optional group activities. For the adventurous, river-loving souls, we have planned an optional riverboat dinner cruise to cap off the conference. For those of you planning to bring children, we are going to offer some special kids' events this year. More information on these activities is included on page 3.

Please note that you are responsible for reserving your own hotel room. The sooner you reserve your room, the better since rooms are limited.

We have had to increase our conference registration fee to \$150/person with no graduated scale for family members. This fee primarily covers the cost of the meals and breaks for each attendee, and a very modest portion will go toward the other expenses for putting on a conference of this scope. Our general operating budget—which is extremely small as a start-up not-for-profit—takes a substantial hit in supplementing the costs associated with these conferences. **For these reasons, we are asking each of you to please consider putting on a fundraiser to help underwrite the substantial costs of this conference.** A fundraiser does not have to be anything elaborate. It can consist of a bake sale, a car wash, a backyard barbecue, or a yard sale. For those of you inclined to fancier tastes, some ideas include a wine-tasting party, a cigar-and-martini night or a golf tournament. Fancy or plain, I think you get my message. **We desperately need money!** If you are willing to host a fundraiser, please contact Alice or me, and we can give you the help you need to carry it off successfully. **Get your registration forms in as soon as possible! I am looking forward to seeing everybody there!**

Lyn Pickel

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.

Make Your Reservations

***To Attend the
3rd Annual Conference of the
Melorheostosis Association***

***July 17-19, 2005
St. Louis, MO***

***Details on Page 2..
Registration Form on Page 3..***

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Central DNA & Tissue Repository Update

At NIH, Dr. Pamela Robey continues to investigate the possibility of establishing a central, neutral repository where patients with melorheostosis, osteopoikilosis and BOS could submit their blood, tissues and DNA for study. Our plan is that researchers from all over the world could apply to the repository for samples, thereby opening the door to vigorous, fair and healthy scientific exploration and competition. **It is possible that we may collect our first round of DNA samples at the upcoming conference!**

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****SPECIAL NOTICE for Patients with Surgery Scheduled****

The best time to collect tissue samples is during surgery. If you are definitely going to have surgery, please consider having tissue samples saved for the repository at the time of your procedure. Anyone interested in donating samples for research should get in touch with Lyn, Kathleen or Alice. Instructions will be provided for sample collection and dissemination.

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'Surgical Consideration' Document

Dr. Jeffrey King has completed a draft of this document. It is currently being reviewed by his colleagues. Once it is in final form, it will be posted on the website. Thank you for your continued work on behalf of melorheostosis, Dr. King!

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Melorheostosis Association
Acting Editor, Alice Albin
(albin@msu.edu: 517-355-7673)*

The highlight of the 2005 Conference will be scientific and medical talks about melorheostosis and research on other diseases which have similar characteristics. The conference will begin on Sunday evening, July 17 with a Welcome Reception. If time permits, there may be a few patient clinical reviews scheduled. Also, we hope to begin drawing DNA specimens for the neutral repository. A formal agenda will be prepared in the next couple of months.

Tentative Schedule

- July 17** **Evening Welcome Reception**
- July 18** **Medical & Scientific Talks**
Optional Children Day Trip(s)
Lunch & Dinner
- July 19** **Patient Clinical Reviews (time permitting)**
DNA Collection for Repository
More Talks
Optional Riverboat Dinner Cruise

Conference Moderator

Dr. Fred Kaplan, Univ. of Pennsylvania

Conference Speakers

- Dr. Francois Glorieux, McGill University, Montreal
- Dr. Jill Helms, Stanford University
- Dr. Jeffrey King, Michigan State University
- Dr. Peter Klein, University of Pennsylvania
- Dr. Laura McCabe, Michigan State University
- Dr. Geert Mortier, Ghent University, Belgium
- Dr. Pam Robey, National Institute of Health
- Dr. Eileen Shore, University of Pennsylvania
- Dr. Michael Whyte, Washington University
- Dr. Howard Worman, Columbia University
- Dr. Michael Zasloff, Georgetown University

Additionally, a speaking invitation has been extended to Dr. Berthold Struk (Franz-Vollhard Clinic for Cardiovascular Medicine and Max Delbrueck Center for Molecular Medicine, Berlin). He will formalize his plans in May, but has replied with a tentative 'yes'.

You can expect unbelievable camaraderie, an exciting and invigorating exchange between all present, and last but not least, great food!

Hope to see you there!!!

Transportation to St. Louis, Missouri

Options include air, car, bus, or Amtrak. For those of you who will be traveling by air, more information will follow on transportation options between the airport and the hotel.

Lodging Details

A block of 40 rooms are being held at the Water Tower Inn on the campus of SLU. The discounted room rate is \$70/night. Rooms accommodate up to 4 people at no extra cost, and include a complementary breakfast. Please phone your reservation in to the:

Watertower Inn
 3545 Lafayette Avenue
 St. Louis, MO 63104
Phone: (314) 977-7500

Mention: Group Reservation #668

Deadline: June 1, 2005

www.slu.edu/events/water_tower_inn.html

Meeting Registration Fees

Meeting registration is \$150 per person. This fee is based on 2004 conference expenses per person and is an estimate of actual costs for the reception, breaks, and meals thru lunch on Tuesday.

Introducing the 2005 Conference Planners

Dr. Robert Fleming, M.D.
SLU School of Medicine

Dr. Fleming is a neonatalist, geneticist, and friend of Lyn Pickel. Thanks to his diligent and gracious efforts St. Louis University has agreed to host the 2005 conference. Dr. Fleming has been following recent melorheostosis articles, has shown much enthusiasm in helping our group plan this conference, and has been generating interest from his colleagues at SLU. He is certain many medical practitioners from SLU and Washington University will attend this conference.

Debbie Tarpe and Madelyn Reilly

Debbie and Madelyn are also friends of Lyn Pickel, and have been volunteering time every week to secure hotel accommodations, meeting facilities, explore catering options, group activities, etc.

Lyn Pickel lynbpickel@earthlink.net
Alice Albin albin@msu.edu

A heartfelt 'thank you' to Dr. Fleming, Debbie and Madelyn!!!

Optional Conference Activities

Children's Activities

Tentatively, we are planning sightseeing trips to the City Museum, the Science Center, the Zoo and the Magic House. Middle and high school students (including Lyn's children!) will be recruited to help chaperone these events. In addition, Donna DeLuca's good friend, Annmarie Fiore, and Lyn's mother, Harriet have magnanimously agreed to help chaperone the children's program. We hope that this component will make the conference fun and memorable for the kids. Please keep in mind, though, that if you bring your children, you are still primarily responsible for supervising them (except for when they are participating in these activities).

These activities will be coordinated on site on a pay-as-you-go basis (admission + transportation). More information will be provided later. At this point we are not planning on collecting funds in advance for these activities.

Riverboat Dinner Cruise

Tuesday, July 19

7:30-9:30 p.m.

The cruise consists of a 2 ½ hour scenic trip down the mighty Mississippi, featuring a lovely dinner and a live Dixieland band.

Cost is \$35 for Adults and \$18 for children ages 3-12. We are collecting for this in advance. If you plan on remaining in St. Louis the evening of July 19, we hope you will join us.

Meal Options:

Prime Rib
Chicken Champagne

2005 CONFERENCE REGISTRATION FORM

Name: _____

Address: _____

Email: _____

____ Registrations @ \$150

Please list others and include age of children:

Please make checks payable to:
Melorheostosis Association

____ Optional Riverboat Dinner (July 19, 7:30-9:30 p.m.)
No. Adults _____ @ \$35
No. Children _____ age 3-12 @ \$18

Specify Riverboat Meal Selection:
Prime Rib: #Adults ____ #Children ____
Champagne Chicken: #Adults ____ #Children ____

Mail check and registration form to:
Mrs. Alice Albin
MSU-DOE Plant Research Laboratory
Michigan State University
East Lansing, MI 48824

\$_____ Amount Enclosed for Riverboat Dinner Cruise

\$_____ Amount Enclosed for Meeting Registration

\$_____ Optional Tax-Deductible Gift (*thank you!*)

Registration Deadline: June 1, 2005

4 New Panel Members

The Board is pleased to announce the addition of 4 new members to the Panel. They are:

Dr. Peter Klein, M.D., Ph.D.

Associate Professor of Medicine &
Cell and Developmental Biology
University of Pennsylvania

Dr. Klein (a co-discoverer of the LEMD3 gene; also called SANE) first described the role of the LEMD3 gene in the bone morphogenetic protein signaling pathway.

Dr. Howard Worman, M.D.

Associate Professor of Medicine
Columbia University

Dr. Worman is responsible for pioneering some of the early work on the LEMD3 gene and identifying its critical role in bone formation.

Dr. Geert Mortier, M.D.

Professor of Medicine
Dept. of Medical Genetics
Ghent Univ. Hospital, Belgium

In October 2004, Dr. Mortier announced the discovery of the mutation in the LEMD3 gene resulting in melorheostosis.

Dr. Laura McCabe, Ph.D.

Associate Prof. of Physiology
and Department of Radiology
College of Osteopathic Medicine
Michigan State University

Dr. Laura McCabe's research interest lies in understanding the regulation of maturation and activity of bone forming osteoblasts. Dr. McCabe also provided invaluable help to Alice Albin in organizing local speakers for the 2004 conference at MSU. Her enthusiasm for our 'cause' is inspiring!

Welcome aboard! We are honored to have each of you!

Current Research Projects

With the foundational work of identifying the composition, mechanisms and role of the LEMD3 gene well underway, several members of our advisory committee have either initiated or are considering studies to understand more precisely what happens to cause melorheostosis and the related disorders of osteopoikilosis and Buschke-Ollendorff Syndrome (BOS). Dr. Geert Mortier is continuing his project and is actively seeking DNA samples from patients with any of the 3 diseases. Dr. Michael Whyte has also launched a research study and is seeking DNA samples from melorheostosis and osteopoikilosis patients.

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Panel Resignations

Ms. Jeannie Peeper has announced that she must withdraw from our Panel effective June 1, 2005. Jeannie has worked closely with the Board on many, many issues related to organizing our Association. Her insight and guidance has been absolutely invaluable, and she will be sorely missed on both a professional and personal level. Our next newsletter will include an article about Jeannie and her work for the IFOP. She is a truly amazing individual. *Thanks for everything, good luck, and our very best wishes to you, Jeannie! You are the best of the best!!!*

Regretfully, Dr. William Cooney, III has also resigned from his position on the panel. We are thankful for the many informational contributions he has made over the past year. While Dr. Cooney's schedule does not allow him to fully participate, it is encouraging that his interest in melorheostosis remains, and he continues to respond to requests for information. This helps us tremendously, and we wish to encourage him to continue participating at whatever level he can. *Many thanks, Dr. Cooney!*

Relevant Scientific Publications

Mortier, GR et al. Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. Nat. Genet. 2004 Nov;36(11):1213-8. Epub 2004 Oct 17.

Lin, F, Morrison, JM, Wu, W, Worman, HJ. MAN1, an integral protein of the inner nuclear membrane, binds Smad2 and Smad3 and antagonizes transforming growth factor-beta signaling. Hum. Mol. Genet. 2005 Feb 1;14(3):437-45. Epub 2004 Dec 15.

Raju GP, Dimova N, Klein PS, Huang HC. SANE, a novel LEM domain protein, regulates bone morphogenetic protein signaling through interaction with Smad1. J Biol Chem. 2003 Jan 3;278(1):428-37. Epub 2002 Oct 21.

Iyer V, McCabe LR, and R Schwartz. CCAAT/Enhancer-Binding Protein-beta has a Role in Osteoblast Proliferation and Differentiation. Experimental Cell Research 295 Entrez PubMed:128-37, 2004.

Other Announcements and Developments...

Donna DeLuca and Alice Albin have finished the gift envelope, and a supply of approximately 900 is available. Lyn Pickel will gladly mail you as many as you need. You need only ask!!

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Alice Albin has set up and is using QuickBooks for Association accounting. Alice continues to work on a draft of the Association Board Policies and Procedures Manual, and the Association Brochure.

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Solicitation approvals have been received for fundraising activities in the following states:

Michigan
New York
Missouri
Massachusetts
Wisconsin
Pennsylvania
Florida
Illinois

If your state is not listed here, and you want to do fundraising, please contact Alice ASAP.

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Did you know? A general 'melorheostosis' search on www.pubmed.com will net 286 articles. Many are case reports, but valuable information can be obtained by reviewing them. Many of the more recent articles are more scientifically-based and research oriented. This is what we want to see!!

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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

Conference Committee – Alice Albin

Scientific/Medical Research Initiatives Committee – Lyn Pickel, Kathleen Harper (Co-Chairs)

On the International Scene...

The U.K. Melorheostosis organization is making great strides towards registering their charity, have developed a wonderful website (www.melo.eu.com), have launched one fundraiser, arranged press coverage, and are planning a large fundraiser this summer. The U.K. group plans to help pay for the U.S. conference. Thank you to the U.K. team!!

How you can help ...

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, and indicate how you would like to help.

Fundraising...

Please think fundraising! We need money to run our organization and sponsor these conferences. Someday we would like to sponsor research, and have a real office with real employees. We are literally squeaking by. Can you help? Here are some ideas:

Open House or Wine Tasting Party
Letter Writing Campaign to Family & Friends
Human Interest Article in Newspaper
Garage Sale
Bake Sale
Golf Outing
Formal Dinner
Church or Work Event
Silent Auction or Raffle
A Race or Walk-A-Thon
Employment/Employee Matching Programs

Celebrity involvement could help us. Does anyone know a celebrity?

In relation to our upcoming conference, does anyone work for a company that would consider sponsoring part of our conference expenses? This conference is estimated to cost \$15-20,000. All gifts to our organization are fully tax deductible.

Please lend a hand in fundraising!!!

Director Contact Information

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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 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