

## Genetics in Osteoporosis: A Short Update

These days everyone seems to be talking about a genetic basis for just about every disease. Indeed, we now know that almost all diseases are somehow influenced, or even caused by, abnormalities in our genes. The same is true of osteoporosis. Bone density, of course, is the measure on which we focus regarding risk of osteoporosis and fractures. This is why bone density has been the focus of genetic studies in osteoporosis. It has been somewhat of a surprise to discover in recent years that nearly 80% of the variation in bone density in a population is determined by our genes, i.e., our inheritance. Thus, one might conclude, “there is not much we can do about our bone density”, and “we must become resigned to our fate if our genes make us have osteoporosis”. Not necessarily true. Most fractures occur late, rather than early, in life and bone loss occurs in everybody late, rather than early, in life. Therefore, if your inherited bone mass is relatively low, you need to consider interventions prior to middle age, and/or prior to menopause, that will prevent age-related bone loss from occurring. So do not despair! Your bone mass may be adequate in young life to prevent fractures, and the amount of bone you once had in young life can be preserved.

It is important to understand that lifestyle does heavily influence the amount of bone you acquire in childhood, and the amount you retain with advancing age. Thus, it is important to eliminate lifestyle factors that increase bone loss as one ages. The most important

ones are lifelong inadequate calcium intake, lifelong vitamin D insufficiency, excessive alcohol consumption, smoking and sedentary lifestyle. This is true regardless of your inherited bone density.

As we learned about secondary causes of osteoporosis in the last newsletter, there is a host of diseases and their treatments that also have a negative influence

on your inherited bone mass. Prominent among them are inflammatory diseases, such as inflammatory arthritis, and allergic diseases that require treatment with cortisone-like drugs. Again, do not despair. We now have methods of countering those problems as well.

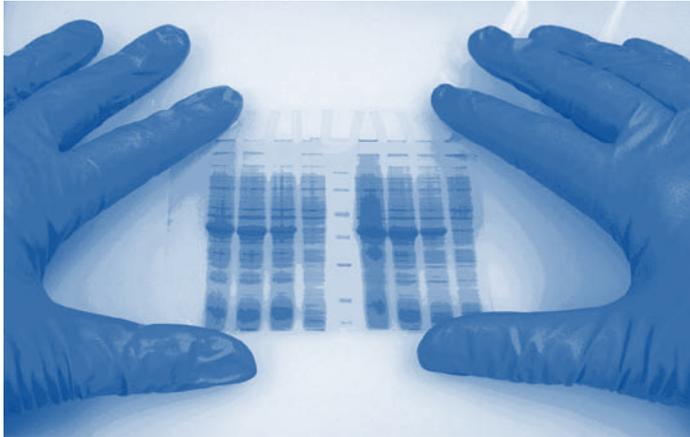


The good news is that we have been able to put genetics to work for us, here is an example. An important discovery was made by research scientists at the Creighton University Osteoporosis Research Center who discovered a gene mutation that resulted in very strong bones. The location of this mutation and its physiology was worked out. This discovery has led to worldwide research in many laboratories attempting to build on this discovery by creating drugs that mimic the effect of the mutation. It is likely that at some time in the future, research based on this discovery will be successful; we hope sooner rather than later.

We in the ORC are studying a large number of genes that influence bone mass, and that influence bone loss. This area of study is being pursued in numerous laboratories all over the world. This promises to

uncover more secrets of nature that can be used in the same manner as the high bone mass mutation that we found.

The mutation that resulted in massive, very strong bones is but one example of the genetic work in progress at the Osteoporosis Research Center. We



have recruited literally thousands of persons from about 700 families to determine **HOW** bone mass is inherited. Each person we have recruited helped the effort by donating his/her DNA, and by allowing us to measure their bone density and record their histories. This has led to dozens of scientific publications describing the genes that determine bone mass and other factors that cause the skeleton to weaken. Now, nearly every human research study we conduct has a genetic component.

For example, we have found in our vitamin D supplementation studies in humans that there is wide variation in vitamin D levels in the population in the face of generally uniform sun exposure. Further, we are finding wide variation in the blood vitamin D levels occurring in response to supplementation with the same dose of vitamin D. We think these factors are influenced by variation in individuals' genes, and we are working to discover the genes responsible for this variation in hopes of learning how to insure that everyone gets sufficient vitamin D, regardless of their genes.

The need for expert genetic research has led to the addition of new faculty in the Osteoporosis Research Center. Three new faculty members have joined us in the past 2 years, Drs. Gary Xiao, Lanjuan Zhao and

Peng Xiao. They are from China, and have trained in the U.S. Drs. Lanjuan Zhao and Peng Xiao did their doctoral and post doctoral training in our genetics laboratory, and agreed to stay on as faculty. They have contributed several prize-winning scientific articles since joining the faculty. Dr. Gary Xiao comes to us from the University of California where he did his post doctoral training. He brings expertise in laboratory technology that we have never had before. We managed to find funds to purchase extensive new laboratory equipment that has opened new avenues of discovery. We are determined to maintain our leading position in genetic research in osteoporosis.

We anticipate that we can make our genes work for us if we conduct the proper research.

## RECENT DONATIONS

### IN HONOR OF

*The Staff of the Osteoporosis Research Center*  
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*Tony and Jeanne Raimondo*  
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## **We need your help!**

A bill has been introduced in the US House of Representatives to create a National Bone Health Program that would have a tremendous impact on the critical problem of osteoporosis. If passed into law, this bill would provide for enhanced education about prevention and treatment of osteoporosis, broader screening for the disorder, and much-needed research.

The mission of the Creighton Osteoporosis Research Center is to serve persons at risk of or suffering from osteoporosis. Please join us in addressing our mission by contacting your Representative and asking him/her to support H. R. 3856.

You can find the name and contact information of your Representative by getting on the computer and going to <https://writerep.house.gov/writerep/welcome.shtml>. You can easily email your Representative from that site.

## **Advocate for Osteoporosis!**

**One in two women age 50 and older and up to one in four men will have an osteoporosis-related fracture in their lifetime.**

Breaking a bone often leads to a downward spiral of pain, disability, deformity and loss of independence and quality of life. Osteoporotic fractures account for \$18 billion in costs and are projected to increase

by 50 percent over the next two decades, reaching \$25.3 billion in 2025.

**Bone density tests are critical to detecting osteoporosis and preventing debilitating and costly fractures before they occur.**

Dual-energy x-ray absorptiometry (DXA) tests can help predict a person's chances of fracturing and help monitor and evaluate a patient's osteoporosis treatment. Since 2006, reimbursement rates for DXA have been reduced by 50 percent, and more cuts are expected in 2010.

**Lower reimbursement rates could impact the health of many Americans with or at risk for osteoporosis.**

Current reimbursement levels do not cover the cost of providing a DXA and many physicians are beginning to reduce or discontinue providing bone density tests. Traveling long distances for a test would be an extreme hardship for those already frail from osteoporosis and low bone density, those living in rural and medically underserved areas, and those with disabilities.

**Minimizing fracture risk through early detection and preventive care far outweigh the costs of DXA testing.**

A 2008 study by Kaiser in Southern California found that increased use of DXA testing and osteoporosis treatment over a five-year period (2002-2006) resulted in a 37 percent reduction in hip fractures and \$30.8 million in savings in a single year in 11 Kaiser health centers.

You can take action. Go to the National Osteoporosis Foundation website at [NOF.org](http://NOF.org), click on "Advocacy" and it will give you the names of your senators and congressmen. At this website you can send letters urging them to cosponsor S. 769/H.R. 1894. Thank you! If you haven't, it's not too late! Send a message to Congress today!

# Opportunities to Participate

*The Creighton University Osteoporosis Research Center is conducting the following studies. If you have any questions, please call 402.280.BONE (2663) or Toll-free 800.368.5097.*

## RESEARCH STUDY FOR 13 AND 14 YEAR OLD GIRLS

This is a one year research study at the Osteoporosis Research Center evaluating the role of dairy consumption on weight management.

Requirements:

- 5 visits
- Painless evaluations
- Monetary stipend

Please contact our nurses at **402.280.4070** for more information.

## GENETIC DETERMINATIONS

The purpose of this study is to identify genes and proteins that may increase the risk of osteoporosis

- Seeking Caucasian women age 50-55
- Free bone density scan (DXA) with interpretation of results for eligible participants
- Stipend available; 2 visits

## ARE YOU DIABETIC?

The Osteoporosis Research Center is currently conducting a study to determine the effects of diabetes on bone health.

Both men & women are eligible for this study. To qualify for participation:

- Must be a type 1 diabetic for at least 3 years.
- Must be between 19 and 50 years of age.
- 3 visits to our center
- 1st visit involves a blood draw and bone density scans.
- Monetary compensation for study visits

## COULD YOU OR SOMEONE YOU KNOW HAVE OSTEOPOROSIS?

If so, consider participating in a research study on osteoporosis (brittle bones).

If you are:

- Female
- Postmenopausal for 2 years or more
- Age 55 - 89

Qualified study participants will receive:

- Study- related physical exam
- Study drug for osteoporosis
- Vitamin D and calcium supplements
- Compensation for travel and some procedures

This study is being conducted at the Creighton University Osteoporosis Research Center.

## WOMEN SMOKERS NEEDED

The Osteoporosis Research Center is currently conducting a genetic study to determine the effect smoking has on bone health. This study involves no medication and is not a stop smoking study. If you qualify you will receive a stipend and bone density test at no cost to you. We are looking for Caucasian women age 30-40 and it involves one visit to our center.

## DO YOU KNOW THAT YOUNG WOMEN CAN BE DIAGNOSED WITH OSTEOPOROSIS?

We are screening for a study involving an in-depth examination of the bone health of premenopausal women.

You might qualify for a bone density test to evaluate your bone health at no cost to you.

We are looking for women who are still having regular cycles between the ages of 20 and 48.

## RESEARCH STUDY FOR WOMEN OVER THE AGE OF 65

The Creighton University Osteoporosis Research Center is currently conducting a study to test whether an investigational drug, given by injection, safely increases the bone mass of the hip when given in addition to the usual care for osteoporosis.

Do you meet the following criteria?

- At least 65 years old and post-menopausal
- Have either never received osteoporosis treatment or have taken a bisphosphonate (such as Actonel®, Fosamax®, or Boniva®) for the past 1-5 years.

Study staff will review additional study criteria with you at the clinic to determine if you are eligible. Study participation lasts 3 years, and volunteers can expect 10 visits to the study doctor for medical evaluations, which include regular bone density tests.

Women often share the gift of their knowledge with generations that follow. Information about osteoporosis and bone health is a gift worth sharing.

**Find out how you can participate in a study that may help new generations fight osteoporosis.**

# PASS IT ON!

## **Are you a woman who has broken a bone within the last five years?**

**If so, you may qualify for a very important study to learn more about underlying causes of osteoporosis in postmenopausal women.**

Dr. Robert Recker, M.D., Director of the ORC, has recently been awarded a grant for \$3.2 million from the National Institutes of Health to determine why women with slightly low or normal bone density suffer osteoporotic fractures. These women may have defects in their bone quality, and there is concern that currently available medications for osteoporosis may not be helpful to them.

Although many women suffer these fractures, it is difficult for us to find these ladies.

Please consider giving us a call if you are a postmenopausal woman who has had a fracture within the last five years that was not associated with an automobile accident. Also, share this information with any of your friends or family who may qualify.

Women who qualify for this study will receive, free of charge, a bone mineral density scan, several tests to assess the underlying causes of osteoporosis and a monetary stipend for their participation.

**For more information, please leave a message on our hotline at 402.280.BONE (2663) or 800.368.5097. Indicate that you are interested in the NIH Bone Quality Study and one of our research staff will get back to you.**

*Thank you for your consideration of this valuable study.*

## **Attention: Persons in rural Nebraska!!**

**We are asking you to spread the word in rural Nebraska about our new study of vitamin D.**

Dr. Joan Lappe, Ph.D., R.N., one of the ORC investigators, has received a grant for \$4.1 million from the National Institutes of Health to determine if vitamin D can reduce the risk of cancer in postmenopausal women. We are randomly sampling the population of postmenopausal women in 14 rural counties and asking them to participate in this study. The counties include: Burt, Butler, Colfax, Cuming, Dodge, Madison, Nance, Platte, Saunders, Sarpy, Stanton, Thurston, Washington and rural Douglas.

A survey company called MSR will be contacting persons in these counties about participation in the study. Unfortunately, we can not take volunteers for this study; participants will be selected.

Please tell your friends, family and neighbors in these counties to look for postcards about the Creighton study in the mail and to pick up the telephone if MSR calls.



Osteoporosis Research Center

Creighton University  
2500 California Plaza  
Omaha, Nebraska 68178



**Address Service Requested**

Phone: 402.280.4470 Fax: 402.280.5173 Website: [osteoporosis.creighton.edu](http://osteoporosis.creighton.edu) e-mail address: [orc@creighton.edu](mailto:orc@creighton.edu)



## THE GIFT OF GIVING

*Consider a donation in honor of a loved one to the Osteoporosis Research Center*

Just mail this form to:

Sister Anne Evers Endowed Research Fund  
Osteoporosis Research Center  
601 North 30<sup>th</sup> Suite 5766  
Omaha NE 68131

A Contribution is enclosed to the Sister Anne Evers Endowed Research Fund

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