

### **New Research:**

We are offering two new research studies in IBMPFD. These are self-report questionnaires, and we would appreciate participation from everyone whose life has been affected by IBMPFD.

**Quality of Life:** This 4-part survey asks about your health, symptoms, diet and exercise. We hope to find ways to improve IBMPFD treatment and how to improve quality of life while coping with the disease.

**The Disclosure Survey:** We want to look at opinions and concerns people have before and after receiving testing for IBMPFD.

Please go to the following websites, or you can email, call or write to Alisara Ateerat, and request a survey to be mailed to you, or to schedule to have one or both administered over the phone.

<http://www.ucihs.uci.edu/pediatrics/drkimonis/index.shtml>

-Ally Ateerat

### **Creating cell lines:**

As part of our continuing studies, we are hoping to create cell lines from as many participants as possible.

**Tissue:** If you are planning on having any type of surgery, please notify us so we can obtain a tiny amount of tissue and skin at the time of the procedure.

**Autopsy:** On a sad note, we learn about loved ones who are going to, or have unfortunately passed away. Where appropriate, obtaining a small amount of tissue at autopsy is extremely beneficial in helping us understand the disease. We can help facilitate this if you are able to contact us, at no charge to yourself. Please contact Dr Kimonis. Tissue obtained at autopsy will be banked at the Maryland Brain and Tissue Bank for future research studies

<http://medschool.umaryland.edu/btbank/gintro.html>.

### **Study Participation:**

As children get older and as families expand, there may be other family members that are interested in participating in our study. Please contact Alisara Ateerat, if you know of other individuals that are interested in participating.

### **Funding:**

Our funding currently is from the NIH and MDA. However, to fully understand IBMPFD, we are always expanding our studies. If you or anyone you know is interested in donating to this important cause please contact us.

Visit [www.ibmpfd.com](http://www.ibmpfd.com):

I wanted to let you know that an IBMPFD family member, Mr. David Sweetman, has set up an internet web site specifically for IBMPFD family members. There is also a wealth of updated information about IBMPFD, including descriptions of ongoing research studies. I regularly visit the web site to make sure it contains the most accurate and up-to-date information about IBMPFD. Nonetheless it would be a beneficial to discuss any medical treatments, or other information you are not sure about, with me or with your local primary doctor before making any important health decisions using this information. I hope you will find this web site informative and useful

-Virginia Kimonis, M.D.

The [www.ibmpfd.com](http://www.ibmpfd.com) web site was established to provide a communication channel for researchers, patients, and care-givers; without the constraints imposed by a formal organization. There are separate sections for a variety of topics, including the following:

1. Research information: includes papers or links, for more detailed knowledge of the mutations and genetics.
2. A section on nutrition: includes supplements that may ease some symptoms
3. Exercise: includes some forms that may extend movement.
4. Aids: includes tools such as wheel chairs, canes, lift seats that can help the patient and care-giver more easily performs various functions.
5. A bulletin board: serves as a place where messages can be exchanged about various topics and ideas for dealing with the disease.

Since this is maintained and funded by me alone, there are no constraints (other than usefulness and civility) imposed on the web site. I am very optimistic about the ability of the human species to accomplish many things, including eventually a treatment then a cure for this mutation.

-David Sweetman

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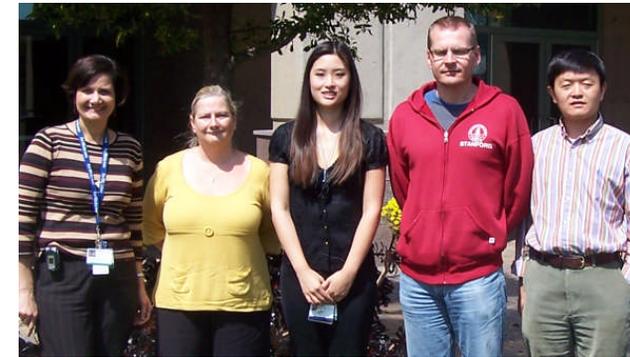
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# *Spring 2008 Newsletter*

## **The Kimonis Laboratory** UC Irvine



*From left to right: Dr. Virginia Kimonis, Dr. June-Anne Gold, Alisara Ateerat, Dr. Jouni Vesa, Dr. Hailing Su*

I am happy to be sending you our second newsletter to update you on our research studies in hereditary inclusion body muscle disease associated with Paget disease of bone +/- frontotemporal dementia. More than 30 families are known with mutations in the VCP, valosin-containing protein. We have developed a mouse model of the most common mutation and hope it will bring us closer to a treatment.

As many of you know I moved to University of California at Irvine in September 2006 and have reestablished the laboratory. I would like to introduce you to Dr Jouni Vesa, an outstanding molecular biologist who is making tremendous progress in understanding the VCP disease. He is joined in the laboratory by Dr Hailing Su, MD, PhD, students and Dr. Mallikarjun Badadani. Dr. Giles Watts continues to work on the bony aspects of VCP disease at Boston Children's Hospital. Alisara Ateerat, M.S genetic counselor requests your help with completing the quality of life questionnaire and will also assist in testing and will study the ethical and social implications of genetic testing in individuals at risk but not manifesting the disease.

We encourage you to participate in the studies of Dr Charles Smith. We are hoping that studies done in our laboratory and that of our collaborators will help alter the course of the disease. We wish you a wonderful year and look forward to hearing from you.

With Warmest Wishes.

-Virginia Kimonis, M.D.