

Multiple Hereditary Exostoses

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FUNTAZIA

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Research Banquet

2007

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* *Magic of Giving!* *

...Be Part of

* *Finding the Cure!!*

* * * * *

Sunday, September 30, 2007

Gargiulo's Restaurant

www.mheresearchfoundation.org



DEAR FAMILY AND FRIENDS,

WELCOME TO “FUNTASIA” 2007! ON BEHALF OF MYSELF, MY FAMILY AND THE MHE RESEARCH FOUNDATION I WOULD LIKE TO THANK ALL OF YOU FOR YOUR CONTINUED SUPPORT AND FAITH IN OUR WORK. WE ARE TRULY DEDICATED TO HELPING VINCENT AND ALL THOSE WHO SUFFER FROM MHE. IT IS ONLY THROUGH RESEARCH, THAT THE DREAMS OF PEOPLE WITH MHE CAN BEGIN TO COME TRUE. WE NEED THEM TO KNOW THAT THERE IS HOPE. THIS IS WHY EACH ONE OF YOU ARE “...PART OF THE MAGIC OF GIVING....AND PART OF THE CURE!” WITHOUT YOUR SUPPORT WE WOULD NOT BE ABLE TO MOVE FORWARD. WE DEEPLY APPRECIATE EVERYTHING THAT YOU HAVE DONE TO HELP MAKE THIS BANQUET A SUCCESS. MAY GOD BLESS ALL OF YOU & YOUR FAMILIES & KEEP YOU IN GOOD HEALTH.

SINCERELY,
SUSAN EATON
(FUNTASIA - CHAIRPERSON)



The MHE Research Foundation's
REACH Research Award



Is presented to
Dominique Stickens, Ph.D.,
Senior Research Biologist,
Merck Research Laboratories, NJ

This Award has been created to show our Foundation's
appreciation to scientist whose past research has had
LONG TERM REACHING affects on the study of
Multiple Hereditary Exostoses.

We hope that this award will inspire researchers around the world to REACH for the CURE and understand the accomplishments of researchers like Dr. Dominique Stickens and future award winners.

Research discoveries are not accomplished over night they take years. Moreover it is these discoveries when put together with others that will one day lead to the discovery of a treatment for Multiple Hereditary Exostoses.

The MHE Research Foundation salutes the recipient of the **REACH Research Award**.

Dr. Stickens has demonstrated his commitment to The MHE Research Foundation time and again and to the research community at large. A noted scientist doing extensive research on MHE, Dr. Stickens has shared his expertise and knowledge whenever asked, and has volunteered to take on the huge task of co-organizing the third International MHE Conference to be held July 2009.

Dr. Stickens was born and raised in Belgium, in a small village near the city of Antwerp. With the exception of his mother (a French teacher), his family consists of many scientists. His grandfather was a medical doctor, his father an engineer and his older brother would eventually become a physicist. The most valuable lesson he received from his parents however was not how to become a scientist, it was a lesson in self-reliance and they let him make his own decisions about his education and career.

In high school he became more and more interested in biology, which soon became the class he most enjoyed. After graduating from high school in 1988 he enrolled at the University of Antwerp in Belgium, studying biology. He completed all the required courses in zoology, botany, chemistry, mathematics, and physics, but he was particularly intrigued by the emerging fields of genetics and biotechnology. It was during his senior year that this interest turned into a true passion, when he joined the lab of Prof. Merregaert to complete his thesis work. During that time he studied how faulty genes contribute to cancer, work that led to his first scientific publication in an international journal. He graduated Magna Cum Laude from University and decided he wanted to pursue this new found passion. His home country however did not provide the opportunities he had hope for and in 1994 he left Belgium to start a PhD program at UT Southwestern Medical Center in Dallas, Texas. It was there that he met Dr. Glen Evans, a prominent scientist in the field of human genetic diseases, who asked him to join his laboratory to work on a disease called hereditary multiple exostoses, a disease Dr. Stickens had never heard of before. What started of as a small project on some unknown disease soon turned into a passionate quest to find out what caused this disease and how that knowledge could help people suffering from this condition. He spend the next ten years, first as a graduate student and later as a postdoctoral fellow at the University of California San Francisco, putting together the genetic pieces of HME puzzle.

Dr. Stickens identified the EXT2 gene, and in so doing, has helped define a new family of putative tumor suppressor genes. Moreover, he went on to characterize that gene for its normal role in development and pursued the question of how a loss of function for this gene product participates in deregulated bone development. Dr. Stickens was able to create a EXT2 knock-out mouse that formed exostoses and thus recreated the human phenotype. The genetically engineered mouse he created has proved to be a very valuable model that provided profound insights into both disease mechanism and basic biological processes. These EXT2 knock-out mice are used by many scientists as a tool for further understanding the underlying causes of MHE. His work and contributions to the understanding of MHE have been presented at several national and international meetings and have led to multiple publications in high-ranking scientific journals.

In April 2005, Merck & Co recruited him as a Senior Research Biologist but he continues to serve as a member of the scientific advisory board of the MHE research foundation and remains dedicated to the field of MHE research.

*The MHE Research Foundation's
Humanitarian and Scientific Achievement Award*



**Is presented to
Yu Yamaguchi, M.D., Ph.D.
Professor Developmental Neurobiology Program,
The Burnham Institute, La Jolla, CA**

This Award has been created to show our Foundation's appreciation to scientists who have shown years of extraordinary commitment to the better understanding of MHE and their years of service to the MHE community.

We hope that children living with MHE can be inspired and understand the accomplishments of Dr. Yu Yamaguchi and the future recipients of this Award. While many people will have to spend their lives overcoming all the physical challenges of MHE, it is their mind, compassion and commitment that can and will pave the way to making their dreams come true. And with this, they will leave their own very special mark in this world and make it a better place for us all.

The MHE Research Foundation salutes the recipient of the **Humanitarian and Scientific Achievement Award**. Dr. Yu Yamaguchi has demonstrated his commitment to The MHE Research Foundation time and again and to the research community at large. A noted scientist doing extensive research on MHE, Dr. Yamaguchi has shared his expertise and knowledge whenever asked, and has volunteered to take on the huge task of co-organizing the third International MHE Conference to be held July 2009.

Dr. Yamaguchi was contacted by our foundation's National Director of Research Sarah Ziegler who spoke with him in detail about all the different symptoms that she had been seeing in people with MHE. She went on to speak with him concerning why she believed that these symptoms were connected to MHE and moreover why Orthopaedic physicians had not connected these symptoms to MHE before, due to the rare nature of this disease. How could any researcher really think about discovering treatment, if they did not know what all symptoms were and how they are connected to Multiple Hereditary Exostoses. Dr. Yamaguchi took on this challenge and over the past 5 years has been a world leader not only in the research he is conducting; but he also brings his discoveries to the scientific community at large. For more than one hundred years MHE was thought of as a bone disease, where by multiple bone tumors formed. Both researcher and physicians now think about MHE to be multifaceted disorder thanks to so much effort given by Dr. Yamaguchi. His research includes neurobiology, bone and cartilage in order to study this disease as a whole with all that this entails. As more of this research emerge both parents and physicians will be able to intervene much earlier and get the child suffering from these symptoms such as learning disabilities the educational resources they need to help overcome these disabilities. The lives of the MHE child is much brighter thanks to not only Dr. Yamaguchi's scientific efforts but also his humanitarian efforts as well.

Dr. Yamaguchi was born at Sendai, Japan, to a family of medical tradition. Both of his parents, both of his grandfathers and great-grand father were all M.Ds. He met his wife Masumi as they both attended Tohoku University School of Medicine and went on to have three children. They had a son Tsubasa, who is now a College senior, a daughter Mai, who just started college this September, and a daughter Shoko, who is now high school senior.

Yu Yamaguchi earned his M.D. from Tohoku University in Japan in 1981, followed by a Ph.D. in 1985. He came to the Burnham Institute for Medical Research for his postdoctoral training. He was appointed to the staff in 1991 where he has continued his research.

Dr. Yamaguchi created a conditional EXT1 knockout mouse model. These conditional EXT1 knockout mice are being used for genetic studies to figure out how the deficiency of EXT1/heparan sulfate causes MHE. These conditional knockout mice, which allow knocking out EXT1 at the site and time of researchers' desire, are very useful for diverse studies on the function of EXT1/heparan sulfate. Dr. Yamaguchi and his lab have been able to distribute these mice to more than 20 laboratories around the world (US, Europe, and Japan) to help studies by other MHE investigators. Using this model system, Dr. Yamaguchi has demonstrated that mutations of EXT1 influence not only bones but also the nervous system. Although frequently ignored in the clinical front, MHE patients tend to have some mental, neurological, and muscular symptoms. Such symptoms include: mild social interaction deficits, heightened sensitivities to sensory stimulation, difficulties to concentrate, and muscle weakness. Dr. Yamaguchi believes these neurological symptoms can be explained by the deficiency of heparan sulfate in nerve cells. Indeed, recent analysis of knockout mouse behavior has revealed that these mice have deficits in certain aspects of learning and the levels of fear/anxiety, as well as alterations in nerve cell wiring. In addition, Dr. Yamaguchi has recently discovered that knockout of EXT1 in embryonic structures that destined to become bones and cartilage causes severe bone abnormalities. These findings have provided us with a new insight into the reason why MHE patients frequently associate a variety of symptoms other than exostoses / osteochondroma formation, and suggest potential novel MHE treatment paradigms.



Pure White Wings

All children are born as pure as small white doves with spirits eager to soar. With nurture and care, over time they spread their wings as they grow. With the warm summer breeze, parents take great joy in watching their kids learn to fly. Flying around the clear blue sky with endless amounts of energy. Darting gracefully between cotton cloud dreams as these little doves fly after one another while playing and chasing their dreams.

*You see our pure white doves were born with the same eager spirit, but also with **BROKEN WINGS**. Wings that are lame, bumpy and crooked, this hampers their ability to fly. Many times they simply watch through a window as others fly to follow their dreams. They can only imagine the feeling of boundless energies of freedom. Imagining what it would be like to dart at will between their cotton cloud dreams, during the warm summer breeze.*

*While their wings maybe broken, their **SPIRITS SOAR**, giving these kids the unmatched strength, courage and determination to overcome and wisdom far beyond their years. It is their **SPIRIT!** Gracing them to find other adventures in life and spotting joy that others over look.*

Their energies are saved for times when it's needed for them to be able to overcome the challenges they face day in and day out. Their strength is used to endure the many surgeries and pain they face, trying to repair broken wings. Hoping that this surgery will be their last.

*The **WINGS OF HOPE** lead the way as we **REACH** for the **CURE!** in order to fix our children's broken wings once and for all. To put an end to a life time of sitting on the side lines, watching others through the window. So they may also one day truly feel all the scenes of freedom others enjoy.*

WHO ARE THESE WINGS OF HOPE? YOU ASK !

THEY ARE EVERYONE OF US!!

*For you see, we are the care takers of the future, so others may one day be able to fly amongst clear warm blue skies of cotton cloud dreams. Just the way life should be. The **MHE / MO / HME RESEARCH** being conducted today around the world will open the **WINDOW** in the future and allow our children not only to **FLY LIKE THE PURE WHITE DOVES THEY ARE!***

BUT SOAR LIKE EALGES!!!

*The **MHE Research Foundation** would like to **THANK** every one of you for becoming one of our **WINGS OF HOPE!!!!***



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Congratulations to Vincent
and all the Eatons.....
Susan, Chris, Danielle, and
even Craig!!!!!!!!!!
...for the great work you
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Your Friends,
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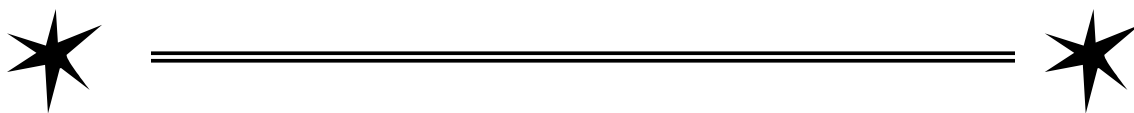




*IN HONOR OF A VERY
SPECIAL YOUNG MAN,
VINCENT EATON
&
FOR CRAIG & SUSAN,
FOR ALL THEY DO
TO HELP OTHERS.*

*Bill & Donna
Guarinello*





Best Wishes

Maureen Moore
Bob, Katie & Bobby Golden





To The Eaton Family,

*We wish you much success with
“Funtasia 2007”.*

*Vincent, you are such a brave
young man.*

*We are happy to be
able to ...*

*Be part of the Magic of Giving,
and to ... Be part of the cure!!!*

*Love,
Fran, Bart, Sallyanne & Victor
Allegretti*





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TO OUR COUSIN VINCENT,



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CONGRATULATIONS ON “FUNTASIA” 2007

Wishing The
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In All Its Efforts To
Raise Awareness Of
This Disease &
To Finding A Cure

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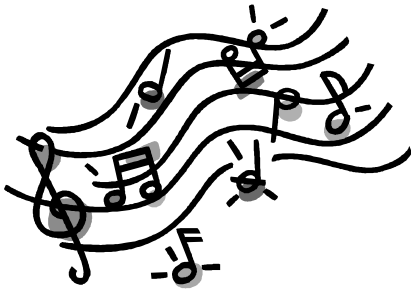
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*To Our Dear Grandson Vincent,
You are one of a kind with your strong personality
and determination. We hope you'll grow up to be
a healthy and successful young man.
Our support and love will always be there for you.
God Bless and Protect you always.*

*Love,
Teta Lily & Sido Shafiq*

*Dear Vincent,

We must all pray that researchers find
a cure to help alleviate the suffering that
this disease causes. MHE has affected
many lives including yours, our grandson
Vincent, whom we love very much.*

*Love,
Grandpa & Grandma Eaton*



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Vincent

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-



To All My Family And Friends,

I would like to thank all of you for supporting "Funtasia 2007". By supporting this Research Banquet, you are supporting myself, and others like me that have to deal with this condition. I am truly blessed to have all of you as my family and friends. I am grateful that you are part of our mission to find a cure for Multiple Hereditary Exostoses (MHE).

Thank you to all those who attended, took a journal ad, made donations or helped with the raffle baskets that made this event a successful one. Whatever your part was I will always remember that all of you helped get me closer to reaching our goals and a cure for MHE.

Love Always,
Vincent





The MHE Research Foundation



Wings of HOPE as we REACH for the
CURE to Multiple Hereditary Exostoses
www.MHEResearchFoundation.org

The MHE Research Foundation is a nonprofit organization for the support of researchers, families and physicians dealing with (MHE) Multiple Hereditary Exostoses / (MO) Multiple Osteochondroma a rare genetic bone disease. The MHE Research Foundation five point mission is to **REACH**, advance & support the following.

RESEARCH, to help fund research programs, so one day a treatment / cure for MHE can be found.

EDUCATION, to provide clinical information, guides to help benefit both families and physicians.

ADVOCACY, to bring awareness about this disease in all areas throughout the world.

CLINICAL, to help provide informational resources to families enabling them to find the medical care they need.

HOPE, is that the research being conducted on MHE, and the information and resources will bring a better quality of life to the families affected by this disease.

Multiple Hereditary Exostoses (“MHE”) is a genetic bone disorder in which benign cartilage-capped tumors (exostoses or osteochondromas) grow from the growth plates of long bones or from the surface of flat bones throughout the body. These exostoses can cause numerous problems, such as: compression of peripheral nerves or blood vessels; irritation of tendons and muscles resulting in pain and loss of motion; skeletal deformity; short stature; limb length discrepancy; chronic pain and fatigue; mobility issues; early onset arthritis; and an increased risk of developing chondrosarcoma. MHE is an autosomal dominant genetic disease and patients have a 50% chance of passing this disorder on to their children.

It is not uncommon for MHE patients to undergo numerous surgical procedures throughout their lives to remove painful or deforming exostoses, to correct limb length discrepancies or to improve range of motion. Surgery, physical therapy and pain management are currently the only options available to MHE patients, but their success varies from patient to patient and many struggle with pain, fatigue and mobility problems throughout their lives.

All Proceeds go to help The MHE Research Foundation in its efforts
to further the understanding of MHE through research and education.

Thank you

The MHE Research Foundation



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