

# HELPING HANDS

FOR FRIENDS, FAMILY, AND SUPPORTERS OF THE CHILDREN'S GAUCHER RESEARCH FUND



## THE ONLY THING INCURABLE IS OUR PASSION.



Valerie Yannias	Victoria Casares	Gregory Austin Macres	Noah Jerome Shaffer	Kyle Herrel	Jared Patrick Ashley	Joseph DeFacci	Andrew Dennis Doran Jr.	James Tyler Cooper	Lauren Marsh	Ryan James Conklin	Cameron Robert Watson	Kristina C.	Madeline Collin	Grant & Garrett	Hannah Colwell	Emma C. Pozzobon Geyer
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*Greg Macres*  
**Gregory Macres**  
Chairman/Founder

Due to a special fund-raising opportunity, we have initiated the writing of this newsletter a few weeks early. At this writing it is April 13, 2005. Eight years ago today we lost our 4-year-old son, little Gregory, as he lay in a Bone Marrow Transplant room at the University of Minnesota battling Type 3 Gaucher Disease. Although the immediate pain has diminished we cannot help but dream of a healthy little Gregory, playing Little League this spring, cuddling with his Mom and Dad on warm summer mornings, and entering Junior High school this coming fall. These thoughts remain dreams, as we focus on the legacy of little Gregory and all the children who have bravely battled this disease. They do have a legacy, and it is one of Commitment, Hope, and a Cure.



*Deborah Macres*  
**Deborah Macres R.N.**  
Founder

### Research

Because of your commitment and support, great progress has been made. A few short years ago very little was known as to how the brain in these children is affected by Gaucher disease. This is now known. In addition, we have believed that the research you have funded would have "overlapping benefits". It is now believed that research on neuronopathic Gaucher disease may help in understanding, and pos-

sibly lead to a cure for some 26 other childhood diseases (Neuronopathic Lysosomal Diseases). With a prevalence of approximately 1:6000 births, the possibilities have expanded beyond one orphan disease. We then ask ourselves if neuronopathic Gaucher disease could be linked to other, more prevalent and well-known neurological disorders. In this newsletter you will read about new, cutting edge research - A link between Gaucher disease, and Parkinson's disease.

### Vlade Divac Joins Hands

As we move down the path toward a cure we work hard to identify creative fund-raising opportunities. Is there a celebrity, an athlete, a well-known figure who would join hands with this effort? Someone we could be proud to associate with – a model of courtesy and respect – a person of character. An athlete who is humble in victory, and gracious in defeat. An athlete who embodies the true meaning of "teamwork". We have found these very qualities in Vlade Divac. Vlade and his wife Ana have joined hands with the Children's Gaucher Research Fund and will work with Coldwell Banker of Sacramento, California, in our Summer 2005 "Pledge For The Cause" fundraising drive.

**“Teamwork is the fuel that allows common people to produce uncommon results.”**

## 100% To Research

So many have done so much. It has truly been teamwork that has allowed the CGRF to make such great progress in a few short years. Our donors – our volunteers who work countless hours – the families who after the loss of a child move forward in a positive direction. We have said it before, "We are humbled by your support". With respect, and a grateful heart we will maintain our commitment. That 100% of every donation will go to medical research.

**A Link Between Parkinson's and Gaucher Disease - See Page 2**

# Coldwell Banker Sacramento and Vlade Divac Team-Up

See Page 2



# Coldwell Banker Sacramento and Vlade Divac Team-Up

It was a shot in the dark on our part, as a call was made to L'Image Restaurant in the Pavilions Shopping Center in Sacramento, an upscale establishment owned by Vlade and Ana Divac. I asked for Ana Divac, and to my surprise she immediately came to the phone. I asked for five minutes to share an idea. Could Coldwell Banker Residential Brokerage and the Divac's work together with the Children's Gaucher Research Fund to raise funds for medical research? After listening she responded, "Because this involves helping children, Vlade will want to know more about it." In the following months we talked about creative ways of working together. Perhaps we were a bit star-struck as we offered ideas that centered the attention on Vlade, after all, he is an NBA star – a celebrity – a Sacramento legend. Time and time again, Ana would say, "Vlade does not want this to be about him. He wants it to be about the charity, he wants it to be about the children." We could elaborate, but instead, we encourage you to read the quotes in this newsletter. Read what others have said. There will then be no need to say more about Vlade and Ana Divac. They are a breath of fresh air in the world of professional sports.

We at the Children's Gaucher Research Fund sincerely thank the team of Coldwell Banker Sacramento and Vlade and Ana Divac for assisting us with our goal:  
**FINDING A CURE.**



## Coldwell Banker's "Pledge For the Cause" Fundraising Drive

All Coldwell Banker real estate agents and employees are welcome to join this effort by soliciting sponsorships from family, friends, neighbors, business associates, companies, etc. All donations are directly donated to the "Children's Gaucher Research Fund" and are fully tax deductible.

"Incredible career. He's really been a positive force and spirit on all the teams he's played for. He's a very special player and person, one that you just don't see in the everyday walk of life."

**Geoff Petrie**  
President of Operations  
Sacramento Kings

"Divac would visit Belgrade in the summer on humanitarian missions, conducting camps and clinics in an effort to help war orphans. He often dipped into his own pockets and played with a heavy heart while his homeland was being bombed."

**Joe Davidson**  
Sacramento Bee

# Is There a Link Between Gaucher



## Mary E. LaMarca

Biologist  
Section on Molecular Neurogenetics  
NSB, National Institute of Mental Health  
National Institutes of Health

Gaucher disease is an example of a "Mendelian" disorder, where defects in a single gene are the primary cause of the disease. Many common human diseases, such as hypertension, diabetes and atherosclerosis, result from interactions between multiple genes and environmental factors such as diet, infections or exposure to toxins. In order to understand the changes in cellular mechanisms that result in these complex, or "multigenic", diseases, researchers can gain information from rare, single gene disorders that share symptoms with more common, complex diseases.

### Gaucher Disease and Parkinsonism

Several new studies have reported evidence for an associa-

tion between Gaucher disease and the development of "parkinsonism". This term refers to neurologic disorders that have many different causes, but share the

classic manifestations of Parkinson disease, such as tremor; stiffness and a shuffling walk. The first hints of a relationship between the two disorders came from scattered case reports of patients with Gaucher disease who developed an early-onset form of parkinsonism. We reported



## Ellen Sidransky M.D.

Chief, Section on Molecular Neurogenetics  
NSB, National Institute of Mental Health and  
MGB, National Human Genome Research Institute  
National Institutes of Health

on a series of 17 such individuals from both Ashkenazi Jewish and diverse ethnic backgrounds. Generally, these patients had relatively mild Gaucher manifestations, with

an average age at diagnosis of 35 years, while their parkinsonian symptoms developed fairly early, with an average age at diagnosis of 48 years. Autopsy evidence from some of these patients showed changes consistent with Parkinson disease, including characteristic structures in brain tissue

known as "Lewy bodies". We also noted that some of these patients had a family history of Parkinson disease, even in relatives who did not have Gaucher disease.

### Parkinson Disease and a Gaucher Gene Mutation

Subsequently, we began a study of brain tissue from individuals with Parkinson disease to investigate how frequently Gaucher (glucocerebrosidase) mutations occur in this population. Mutations in the glucocerebrosidase gene are rare; the carrier frequency for the two most common mutations, N370S and c.84insG, in people of Ashkenazi Jewish descent is about 6%, and is only about 0.4-0.8% in the

**"Several new studies have reported evidence for an association between Gaucher disease and the development of "parkinsonism."**

## Celebration Party - July 28th

On July 28, 2005, there will be a Coldwell Banker Celebration party at the Divac's restaurant, L'Image, located in the Pavilions Shopping Center in Sacramento. Vlade and Ana will join us on a beautiful summer evening for food, music, and fun. To attend the party, you may either purchase a ticket or qualify by:

- Any individual who collects sponsorships totaling \$200
- Every \$200 increment allows the individual to bring a guest
- Any individual who donates \$200 or more also will receive a ticket.
- Example: \$600 qualifies for three attendees (1 ticket / 2 guest tickets)
- All donations must be received by June 20, 2005

## Grand Prize Dinner - July 21st

The grand prize is a private dinner for eight, with Vlade and Ana Divac. The dinner will take place on July 21, 2005, at L'Image restaurant. Who will win the grand prize?

- Whoever raises the highest dollar amount of sponsorships.
- If an individual wins, they and seven guests will attend dinner.
- Teams of four can be created and their collective sponsorships will be counted.
- If a team of four wins, they and four guests will attend dinner.
- To better the chances of winning, teams of four are encouraged.



"A Center of Compassion. Sacramento King Vlade Divac is famous for his assists—both on and off the court. He has a soft touch with his jumper, and a soft heart when it comes to social justice and philanthropy."

**Melinda Welsh**  
Sacramento News & Review

"Who has been more accommodating with autographs, photos and media requests? Who more generously devoted energies to local charities?"

**Ailene Voisin**  
Sacramento Bee

# Disease and Parkinson Disease?

57 subjects with pathologically confirmed Parkinson disease, and from 44 age-matched controls without pathological evidence of Parkinson disease, from five different brain banks around the USA. (Brain banks store tissues from individuals who donate their bodies for research purposes.) To our surprise, we discovered that Gaucher mutations were detected more often than expected in the Parkinson samples. Direct sequencing of the entire glucocerebrosidase gene revealed that eight (14%) harbored at least one Gaucher mutation, while none were found in the controls. The individuals with the Gaucher mutations tended to be among the younger subjects screened. These findings suggested that mutations in the glucocerebrosidase gene, even in carriers, might be a significant inherited risk factor for the development of parkinsonism.

When we speak of a risk factor, it is very different from identifying a gene or gene defect that is directly responsible for the disease. Risk factors can be environmental, like smoking, diet or exposure to toxins, or genetic. As mentioned above, disorders such as Parkinson disease, diabetes or asthma are considered complex

because interactions between multiple genes and environmental factors determine whether an individual will develop the disease. If a person inherits one particular gene defect that is a risk factor, it does not mean that he or she will be affected. However, if the person inherits several contrib-

uting genes or is exposed to environmental factors, he or she may be more at risk to develop the disease in question.

### **Supporting Evidence**

In the past few months, several groups have reported findings that show an increased frequency of Gaucher mutations in patients with parkinsonism. In a series of

a New York City clinic, however, 160 Ashkenazi Jewish probands with Parkinson disease and 92 age-matched controls of Jewish ancestry were screened for just the N370S mutation. The authors found 17 probands (10.7%) with N370S, as compared to 4.3% of controls. These results differ remarkably from the frequency described in the Israeli cohort, so more extensive, larger-scale studies are needed in the Ashkenazi population to establish the true frequency of this finding.

A group from Toronto screened for seven glucocerebrosidase mutations in 88 unrelated Caucasian subjects of Canadian origin with clinically diagnosed Parkinson disease, selected for an early age of onset or a positive family history. These subjects were compared to 122 clinically screened controls. The authors identified mutations in 5.6% of the co

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**"For the medical community, awareness of this association is important to help better understand the possible relationship between Gaucher and Parkinson disease."**

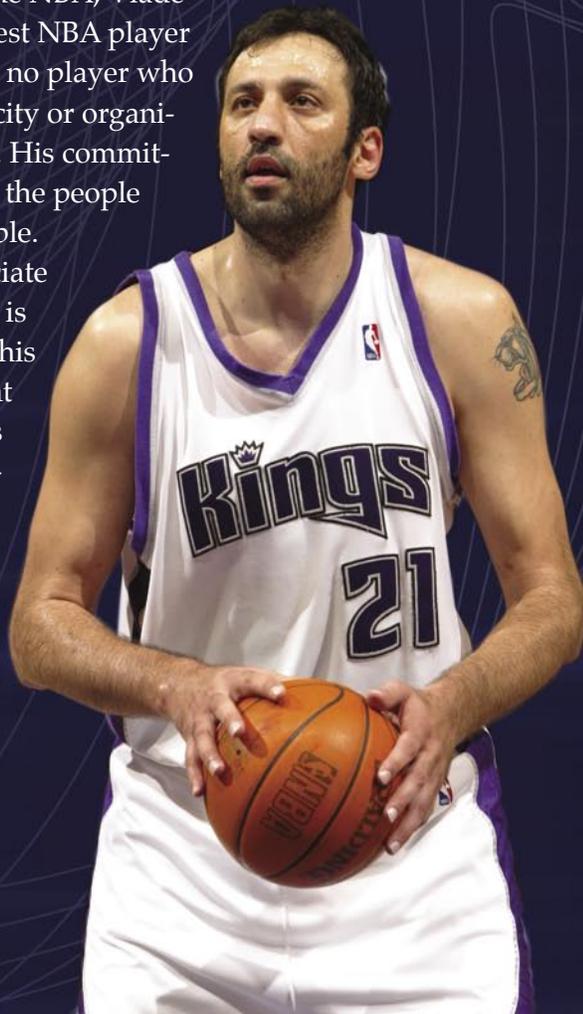
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99 Ashkenazi Jewish patients with classic Parkinson disease seen at a clinic in Northern Israel, 31 (31.3%) were found to carry the glucocerebrosidase mutations N370S or c.84insG. This frequency was at least five-fold higher than observed in the study's two control groups. In a report from

“In my 17 years of association with the Sacramento Kings and the NBA, Vlade Divac is by far the classiest NBA player I have ever met. There is no player who has meant as much to a city or organization than Vlade Divac. His commitment to Sacramento and the people of Sacramento is incredible. For Vlade Divac to associate himself with this charity is very special. For he and his wife Ana to be significant part of this charity, it has to have incredible meaning to them.”



**- Grant Napear**  
Sacramento Kings Play-By-Play  
Announcer and Host of the  
Grant Napear Show



- Vlade Divac (pronounced VLAH-day DEE-vats)
- Height 7' 1"
- From Serbia-Montenegro
- 1985 - Yugoslavian Junior Team - World University Games Gold Medal
- 1988 — Seoul Olympics - Yugoslavian National Team - Silver Medal
- 1989 — First Round NBA Draft — Los Angeles Lakers — 7 seasons
- 1990 - NBA All-Rookie First Team
- 1996 - Olympics - Yugoslavian National Team - Silver Medal
- 1999 - Joined the Sacramento Kings - 6 Seasons
- 2000 - J. Walter Kennedy Citizenship Award for “exemplary community service” by an NBA player
- 2001 - Western Conference All-Star Team
- 2002 — Goodwill Ambassador for the United Nations’
- Vlade is one of only three players in NBA history to amass 13,000 points (13,364)
- Founded the “Divac Fund” through the St. John Foundation to help raise money for children affected by the war in Yugoslavia
- Vlade and his wife, Ana, have two sons (Luka and Matia) and a daughter (Petra) whom they adopted from an orphanage in Belgrade, Yugoslavia

# VLAD DIVAC

hort with Parkinson disease, as compared to 0.8% of the control group. These independent studies, despite their differences in design and patient population, suggest that Gaucher mutations are found in subjects with parkinsonism at a frequency higher than expected carrier frequencies.

### **Then We Approached It Another Way**

Using a totally different approach, we asked the families of our patients with Gaucher disease about relatives with parkinsonism. We identified ten families where carriers of Gaucher mutations, usually a parent or grandparent of the Gaucher patient, developed parkinsonian manifestations. As some of these families were self-referred, it is still too early to establish how common this finding is among all families of patients with Gaucher disease.

### **Awareness Is Important**

For the medical community,

awareness of this association is important to help better understand the possible relationship between Gaucher and Parkinson disease. Patients with Gaucher disease should be specifically questioned regarding a family history of tremor or dementia and, likewise, subjects with parkinsonism might

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**“Researchers hope that understanding how defects in glucocerebrosidase might affect the symptoms of Parkinson disease will lead to the development of more effective treatments for both disorders.”**

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be asked whether any relatives have Gaucher disease.

As a patient, if you have a relative with Parkinson-related symptoms, it would be worthwhile to inform your doctor. However, it is not necessary to live in fear of developing Parkinson disease - this is still a very rare finding. Based upon

the clinical histories of thousands of patients with Gaucher disease, it is evident that the vast majority never develop Parkinson disease. Likewise, most Gaucher carriers do not have Parkinson disease, and, even in families where it is seen, some carriers develop it and others do not.

It is also important to remember that Parkinson disease by itself is a very common disorder, present in 1% of the population, and in as many as 3% of older Americans. Currently, especially in light of the different frequencies reported in these studies, it is best to consider that mutations in the glucocer-

ebrosidase gene are just one of a multitude of potential risk factors for Parkinson disease to which all of us are exposed.

### **What Does All This Mean?**

Research into the possible links between Gaucher disease and Parkinson disease is continuing in multiple laboratories. Some are looking at the frequency of Gaucher mutations in groups of Parkinson patients from different ethnic backgrounds. Others are studying the ways in which alterations in the glucocerebrosidase protein could affect cells in the brain and contribute to the changes seen in parkinsonism. Researchers hope that understanding how defects in glucocerebrosidase might affect the symptoms of Parkinson disease will lead to the development of more effective treatments for both disorders.

## Robert Tabares Steps Up – And So Do The Kids



In the last newsletter we told you about the students at Florin High School in Sacramento, California, who organized an Awake-A-Thon and raised \$4,000 for CGRF research. When we make a mistake we always print a correction. When the final tally was in, they had raised \$8,000.

We would like to recognize and sincerely thank Robert Tabares, (KEY Club faculty advisor) for his tireless efforts. He helped to organize the event, motivate the students, and with his guidance 2x the original goal was raised for medical research.

KEY (Kiwanis Educating Youth) Club was founded in 1925 and plays an important role in positively influencing our youth. The KEY club incorporates a values-based leadership curriculum that emphasizes personal and character development, inclusiveness, and caring. Student officers operate the organization at the club, district, and international levels. Internationally, there are nearly 250,000 Key Club members that comprise close to 5,000 clubs. At Florin High School, there are over 200 members. We thank these students for choosing the Children's Gaucher Research Fund as their "charity of choice".

## Cavitt Junior High School Gives A Thumbs Up



The student government class at Cavitt Junior High School in Granite Bay, California, was looking to create an all-school event that would rally the student body to become involved in raising money for a local charity. Ashley Macres, a member of the student government, suggested a "Cavitt Awake-A-Thon" modeled after the successful Awake-A-Thon that took place last November at Florin High School (SEE "Robert Tabares Steps Up – And So Do The Kids" - above). The student Government class then chose the Children's Gaucher Research Fund as their "charity of choice." We would like to thank all of the students who represent Cavitt Junior High School student government. In addition, we would like to thank Craig Cook (Student Government Teacher) for his support.

## Col(s) Kondi Wong M.D. USAF MC joins the Scientific Advisory Board

Dr. Kondi Wong received his BS from Stanford University in 1982, where he worked in lipid biochemistry and received a research stipend from the American Gastrointestinal Association in 1981. He received his MD in 1988 from the F. Ed-



ward Hebert School of Medicine of the Uniformed Services University of Health Sciences (USUHS) in Bethesda, Md. He completed his residency in 1993 with one year of Internal Medicine and four years in Anatomic and Clinical Pathology at Wilford Hall Medical Center in San Antonio, Texas.

After his residency, he accepted a combined two-year research scholarship and Neuropathology fellowship with Drs. Prusiner, DeArmond, and Davis at the University of California, San Francisco. At UCSF, Dr. Wong's research efforts in Prion and Neuromuscular diseases with Drs. Prusiner and DeArmond were awarded with the 1994 Edwin Boldrey Award for Excellence in Basic Research in the Neurological Sciences.

He came to the AFIP (Air Force Institute of Pathology) as a staff pathologist in the Department of Neuropathology in 1995 and became Chief of the Division of Neuromuscular Pathology in 1998. While at the AFIP, he developed the AFIP Muscle Disorders Course and was awarded the Moore Award for clinicopathologic research from the American Association of Neuropathologists, the AFIP John Hill Brinton award for young investigators research, the Walter Reed Army Medical Center Medallion presented by General Timboe, and Dept. of Defense invention award for patent innovation. After 8 years at the AFIP, Dr. Wong became the Division Chief of Neuropathology at Wilford Hall Medical Center in San Antonio, Texas in July of 2003 and serves as consultant Neuropathologist and as Professor of Pathology at the University of Texas, Health Science Center, San Antonio in an adjunct position.

Dr. Wong is a member of the Scientific Advisory Board for the Children's Gaucher Research Fund.

## In Loving Memory of Shane Michael Hensley



### “Our little ‘Boo Bear”

November 30, 2003 to October 8, 2004  
Fort Pierce  
Florida

## In Loving Memory of Jordan Michael Coles



### “Our Angel In Heaven”

May 25, 1990 to August 11, 2004  
Winnipeg Manitoba  
Canada

## In Loving Memory of Jessica Trautman



### “Our little noodlehead is now at peace”

April 26, 2002 to July 4, 2004  
Virginia Beach  
Virginia

## 100% To Research

You need to know:

1. The CCRF is a **legitimate** IRS approved 501 c3 non-profit organization.
2. **100% of every donation** goes to medical research.
3. We **do not** hire professional fundraising companies who keep 50% of donated funds.
4. We have talented volunteers who **donate** their time and talent for a variety of our needs.
5. All administrative costs are paid for by the **founders**.

Simply put:

If you send your hard earned dollars - **It ALL goes to medical research.**

Visit our web site at:  
[www.childrensgaucher.org](http://www.childrensgaucher.org)  
All family stories can be  
read on the website.

**Contributions Payable To:**  
Children's Gaucher Research Fund  
P.O. Box 2123  
Granite Bay, California 95746-2123

## Children's Gaucher Research Fund



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