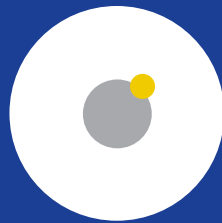


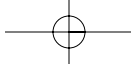
\$5 Million Campaign



SIGHT · SOUND · SPIRIT

EYE ON JACOB

Funding Research for Usher Syndrome



SIGHT · SOUND · SPIRIT
EYE ON JACOB
Funding Research for Usher Syndrome

MISSION STATEMENT

The primary function of Eye on Jacob is to fund research for treatment and, hopefully, a cure for Usher Syndrome. The secondary mission is to establish support groups for patients and families of patients with Usher Syndrome.

BOARD OF DIRECTORS

Sonia Desormeaux, *Board Chair and Founder*

Kent Desormeaux

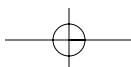
Thomas Anthony Dowd, III

Bruce Hochman

Danielle Merollo

Judge Eddie Sapir

Kelly Wietsma



MESSAGE FROM SONIA DESORMEAUX Board Chair and Founder Eye on Jacob Foundation



Kent and I have two sons, Joshua who is 16 and Jacob who is 9. Joshua is a normal, healthy teenager, and Jacob, as we have discovered, has Usher Syndrome, a very complicated genetic disease that affects children from birth. Usher Syndrome has characteristics very similar to many other genetic diseases, making it very difficult to diagnose.

Jacob was born deaf and has had to overcome many obstacles in his nine years including seventeen surgeries, acute balance issues, and night blindness. One of the greatest challenges of Usher Syndrome patients is the loss of sight in their early adulthood. Though it was difficult, I told Jacob that his night blindness would soon become daytime blindness as well. His response was “Mom, please don’t cry. You always find me the best doctors and I know you won’t stop until you find someone to fix my eyes.”

The purpose of the Eye on Jacob Foundation is to fund research for treatment and hopefully find a cure for Usher Syndrome plus offset the cost of support groups for Usher patients and their families. Hopefully, there will be many positive discoveries made on the journey toward a cure...maybe the first will be a more accurate method for diagnosing the disease...perhaps the second will be an effective treatment for Usher Syndrome.

We know that research takes time and we are not counting on this program to help our Jacob. We are counting on this program to help the tens of thousands of newly diagnosed Usher Syndrome children and their families, and those yet to be diagnosed. With your help, new and better therapies and, hopefully a cure, can be found, and life for Usher Syndrome patients can be made as normal as possible.

ABOUT USHER SYNDROME

Usher Syndrome is an extremely complicated genetic disease to diagnose because it has many of the same characteristics of other genetic diseases...and often it is misdiagnosed. Below is a brief explanation of Usher Syndrome. It is apparent that although a great deal is known about the disease, there is still much more to be learned and research to be done to find more effective treatment and ultimately a cure for Usher Syndrome.

What is Usher Syndrome?

Usher Syndrome is the most common condition that affects both hearing and vision. The major symptoms of Usher Syndrome are hearing loss and an eye disorder called retinitis pigmentosa, or RP. RP causes night-blindness and a loss of peripheral vision (side vision) through the progressive degeneration of the retina, the light-sensitive tissue at the back of the eye which is crucial for vision. As RP progresses, the field of vision narrows until only the ability to see straight ahead remains. Many people with Usher Syndrome also have severe balance problems.

There are three clinical types of Usher Syndrome: type 1, type 2, and type 3. In the United States, types 1 and 2 are the most common types. Together, they account for approximately 90 to 95 percent of all cases of children who have Usher Syndrome.

What are the characteristics of the three types of Usher Syndrome?



Type 1

Children with type 1 Usher Syndrome are profoundly deaf at birth and have severe balance problems. Because of the balance problems associated with type 1 Usher Syndrome, children with this disorder are slow to sit without support and typically don't walk independently before they are 18 months old. These children usually begin to develop vision problems in early childhood, almost always by the time they reach age 10. Vision problems most often begin with difficulty seeing at night, but tend to progress rapidly until the person is completely blind.



Type 2

Children with type 2 Usher Syndrome are born with moderate to severe hearing loss and normal balance. Although the severity of hearing loss varies, most of these children can benefit from hearing aids and can communicate orally. The vision problems in type 2 Usher Syndrome tend to progress more slowly than those in type 1, with the onset of RP often not apparent until the teens.

Type 3

Children with type 3 Usher Syndrome have normal hearing at birth. Although most children with the disorder have normal to near-normal balance, some may develop balance problems later on. Hearing and sight worsen over time, but the rate at which they decline can vary from person to person, even within the same family. A person with type 3 Usher Syndrome may develop hearing loss by the teens, and he or she will usually require hearing aids by mid- to late adulthood. Night blindness usually begins sometime during puberty. Blind spots appear by the late teens to early adulthood, and, by mid-adulthood, the person is usually legally blind.

What causes Usher Syndrome?

Usher Syndrome is inherited from parents to their children through genes. Every person inherits two copies of each gene, one from each parent. Sometimes genes are altered, or mutated. Mutated genes may cause cells to act differently than expected.



Usher Syndrome is inherited as an autosomal recessive trait which means that both males and females can have the disorder and can pass it along to a child. To have Usher Syndrome, a person must receive a mutated form of the Usher Syndrome gene from each parent. If a child has a mutation in one Usher Syndrome gene but the other gene is normal, he or she is predicted to have normal vision and hearing. People with a mutation in a gene that can cause an autosomal recessive disorder can carry the gene with a mutation, but show no symptoms of the disorder. If both parents are carriers of a mutated gene for Usher Syndrome, they will have a one-in-four chance of having a child with Usher Syndrome with each birth.

Usually, parents who have normal hearing and vision do not know if they are carriers of an Usher Syndrome gene mutation. Currently, it is not possible to determine whether a person who does not have a family history of Usher Syndrome is a carrier.

How is Usher Syndrome diagnosed?

Because Usher Syndrome affects hearing, balance, and vision, diagnosis of the disorder usually includes the evaluation of all three senses. Evaluation of the eyes may include a visual field test to measure a person's peripheral vision, an electroretinogram (ERG) to measure the electrical response of the eye's light-sensitive cells, and a retinal examination to observe the retina and other structures in the back of the eye. A hearing (audiologic) evaluation measures how loud sounds at a range of frequencies need to be before a person can hear them. An electronystagmogram (ENG) measures involuntary eye movements that could signify a balance problem.

Early diagnosis of Usher Syndrome is very important. The earlier that parents know if their child has Usher Syndrome, the sooner that child can begin special educational training programs to manage the loss of hearing and vision.

Is genetic testing for Usher Syndrome available?

So far, 11 genetic loci (a segment of chromosome on which a certain gene is located) have been found to cause Usher Syndrome, and nine genes have been pinpointed that cause the disorder.

With so many possible genes involved in Usher Syndrome, genetic tests for the disorder are not conducted on a widespread basis. Diagnosis of Usher Syndrome is usually performed through hearing, balance, and vision tests. Genetic testing for a few of the identified genes is clinically available. Genetic testing for additional Usher Syndrome genes may be available through clinical research studies.

How is Usher Syndrome treated?

Currently, there is no cure for Usher Syndrome. The best treatment involves early identification so that educational programs can begin as soon as possible.

THE RESEARCH TEAM SELECTION PROCESS

Mrs. Desormeaux faced the challenge of finding a physician to head the research team for Eye on Jacob. The Silberstein Group, a highly successful fundraising firm in Houston, Texas, conducted extensive research to find the best physicians, scientists and institutions in the United States doing work on degenerative and genetic eye diseases, especially those involving Usher Syndrome. After a great deal of investigation, the choices were narrowed down to six. Next Mrs. Desormeaux needed to meet the doctors, scientists and staff that would be involved with the research, as well as tour the facilities and laboratories where it would all take place. The Silberstein Group arranged Mrs. Desormeaux's meetings with each of them. Her journey began in late September and lasted through early November of 2008.

Mrs. Desormeaux's first visit was with Dr. Ray Alford, an Associate Professor and Coordinator of Academic and Scientific Program Development at Baylor College of Medicine at the Texas Medical Center in Houston, Texas. She also had the opportunity to meet with Dr. Bobby Alford, Chair of the Bobby R. Alford Department of Otolaryngology-Head and Neck Surgery at Baylor. Dr. Alford was the first person that Mrs. Desormeaux met who gave her hope since she had done research on retinitis pigmentosa. It was positive for her to hear that someone was already working on a cure for sight and vision loss.

Arrangements had also been made for Mrs. Desormeaux to visit Texas Children's Hospital, one of the nation's top children's hospitals, while at the Texas Medical Center. There she met Dr. Richard Lewis of the Department of Ophthalmology and the impressive group of physicians and scientists he had assembled. Among those attending the meeting were a scientist responsible for identifying the type 2 Usher Syndrome gene, a specialist involved in cochlear implants, a pediatric neuro-ophthalmologist, a physician specializing in developmental biology and regeneration of the otic placode, an ophthalmologist whose research has involved the mapping and cloning of genes of hereditary impairment and Usher Syndrome, a pediatric geneticist, and a scientist whose work includes studies involving cochlear hair cells. After this meeting and their marvelous presentation, Mrs. Desormeaux again felt hopeful.



Next Mrs. Desormeaux traveled to Berkeley, California where she met with Dr. John Flannery, professor of Vision Science in the School of Optometry and the Department of Molecular and Cell Biology at the University of California, Berkeley. She went there already aware that the studies done in Dr. Flannery's clinic exclusively deal with type 3 Usher Syndrome. However, while there, she witnessed science and research first hand. She observed tests being performed on mice, saw how the results were interpreted, how genes were mutated, and how sight degenerates. She watched as they froze an eyeball and shaved it and then saw the various layers of the eye. Dr. Flannery and his team have made incredible advances in their research and the work they are doing is remarkable. Her meeting with Dr. Flannery was extremely educational and made her feel even more hopeful that a cure for Usher Syndrome could be found.

Mrs. Desormeaux then went to Iowa City, Iowa to meet with Dr. Edwin Stone, a specialist in genetic eye diseases at the University of Iowa. She had contacted Dr. William Kimberling, Director of the National Center for the Study and Treatment of Usher Syndrome at Boys Town National Research Hospital in Omaha, Nebraska, another physician under consideration on her list, to finalize arrangements to meet with him at a later date. When she mentioned to him that she was planning to meet with Dr. Stone, he offered to meet her in Iowa. Dr. Kimberling met Mrs. Desormeaux and personally took her on a tour of the university and the labs where Mrs. Desormeaux was able to see how they separate genes and DNA, how they identify mutations and various tests that can be done. She was thrilled to learn about the new tests that have been developed to determine the type of Usher Syndrome a patient has from Dr. Kimberling. These tests are considerably less expensive than previous tests (\$35 instead of the previous \$1400) making testing affordable to more patients and therefore enabling more people to be tested for research. Mrs. Desormeaux felt very positive when she left there, and especially excited about reaching out to patients diagnosed with Usher Syndrome to test their DNA.

The final meeting took place in Philadelphia, Pennsylvania at the University of Pennsylvania School of Medicine where Mrs. Desormeaux met with Dr. Jean Bennett, an ophthalmologist specializing in Cell and Developmental Biology whose research centers around the molecular genetics of inherited retinal degenerations. Dr. Bennett took Mrs. Desormeaux on a tour of her laboratory where she was able to see some of the research in progress and introduced her to the scientists and researchers working there. Dr. Bennett and her team had successfully treated a blind dog with gene therapy, that has been seeing well since then with just a single treatment. The first step toward the development of this treatment began with the discovery of the mutated gene which was then cloned and implanted in one eye, restoring the dog's vision in that eye. Clinical trials using this procedure on humans have already begun. Mrs. Desormeaux found this group to be far ahead of anyone else doing gene therapy. They have developed a sterile laboratory where an elaborate machine clones and grows the genes and implants to be used in replacement. Mrs. Desormeaux was extremely impressed by the research being done here as well as with the physicians, the researchers and the entire staff that she met and left there feeling that this might be the perfect fit for Eye on Jacob research funding.

After meeting all six candidates, Mrs. Desormeaux now faced the daunting task of selecting which one would receive the Eye on Jacob funding. She carefully considered each candidate, their research and how each could impact the mission of Eye on Jacob. Meeting these physicians, their teams of scientists and researchers, and observing some of the incredible progress they have already made in restoring sight reinforced Mrs. Desormeaux's hope that a better treatment and hopefully even a cure for Usher Syndrome is possible. Although the decision was difficult, Mrs. Desormeaux chose Dr. Bennett of the University of Pennsylvania. Considering the extraordinary advances that Dr. Bennett and her colleagues have made using gene therapy, the clinical trials that they are doing, the state of the art laboratory and the incredible support they have from The Children's Hospital of Philadelphia, Mrs. Desormeaux feels extremely confident about her choice.

THE UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE-
CHILDREN'S HOSPITAL OF PHILADELPHIA
Eye on Jacob Program for Usher Syndrome

We propose to establish a comprehensive clinical treatment, resource and research program for individuals with Usher Syndrome. This program, supported by The Eye on Jacob Fund for Usher Syndrome at The Children's Hospital of Philadelphia (CHOP) and the Eye on Jacob Fund for Usher Syndrome at the University of Pennsylvania (PENN) School of Medicine will combine the efforts of physicians and scientists with primary appointments at both CHOP and at PENN.

The goal of this program will be to diagnose and evaluate the vision/hearing deficits, to improve our understanding of and to develop therapies for Usher Syndrome, and to provide support to affected patients and their families. A unique aspect of this proposal is the fact that the clinical services are provided at a Children's Hospital. Since Usher Syndrome is a congenital disease, the visits will take place in an atmosphere where both pediatric and adult patients can be assisted.

The long-term goals of the Eye on Jacob Program are to develop therapies for Usher Syndrome. These goals will be pursued via a combination of clinical and lab-based research. This will be a truly "translational" initiative which is multidisciplinary in nature. This Program will rely on close interactions between physicians who characterize the diseases, and scientists who develop the methods with which to document and measure disease progression and/or the effects of treatment thereof. The involvement of experts in clinical trial design and implementation will provide the capacity to perform both pre-clinical and clinical testing of promising treatments.

Further, this Program will serve as a resource for patients and their families by facilitating the diagnostic and treatment processes and by facilitating interactions with other families and professionals. This team approach – involving patients, physicians and scientists – will minimize the burden of this disease, and it will also expedite the development of innovative treatments.



The Eye on Jacob Program will consist of three different components:

- 1) a comprehensive clinical evaluation service for patients with Usher Syndrome. This service will carry out genetic diagnoses, perform ophthalmologic testing, perform hearing and balance evaluations and offer treatments (for example, cochlear implantation, language acquisition therapy) where they may exist.
- 2) a team carrying out translational research on Usher Syndrome. The team will initially focus on one form of Usher Syndrome –USH1C. The goal of the translational research team will be to obtain the appropriate safety and efficacy data as quickly as possible so that a clinical trial can be initiated aiming to reverse the development of blindness in USH1C. Approaches for treating deafness/vestibular problems in this disease will be evaluated simultaneously. These may be tested in a future clinical trial. It is expected that it will be possible to extrapolate any success in developing treatments for the blindness (and/or deafness/vestibular dysfunction) in USH1C to other forms of forms of Usher Syndrome.
- 3) support services for Usher Syndrome patients and their families. These services will include support in daily living (mobility training, visual aides, communication therapy) as well as emotional and structural support for families (including assistance with family, school and occupation issues).

Our intention is to build on our previous experiences in developing treatments for inherited blindness to streamline a gene-based treatment for Usher Syndrome. In doing so, we will share our progress with other clinical and research centers in the United States and around the world so that the maximum number of people will benefit.



PROGRAM/MODULE DIRECTORS

Director of the Eye on Jacob Initiative; Director of the Translational Research Program

Jean Bennett, MD, PhD

Jean Bennett is a pioneer in retinal gene therapy and internationally recognized for her work in this field. She has developed gene transfer approaches to treat retinal degenerative and ocular neovascular diseases, to elucidate retinal differentiation pathways and to identify pathogenetic mechanisms which lead to blindness. She has successfully delivered corrective genes to retinas of animal models that suffer from loss-of-function mutations. This led to the first report of success in slowing retinal degeneration in an animal model of retinitis pigmentosa (RP). Since then, she has been developing treatments for other forms of retinal degeneration (including additional models of RP, Stargardt Disease, lysosomal storage disease, and congenital blindness). In addition, her laboratory has been developing gene transfer approaches to treat syndromic diseases resulting in vision loss, such as Usher Syndrome. Her research, conducted at Penn over the past 16 years, has established the scientific underpinnings which made possible to test the first potential definitive retinal gene therapy treatment for patients with blinding retinal degenerations. This trial, which evaluates the safety and efficacy of gene augmentation for a disease called Leber Congenital Amaurosis, was initiated in the fall of 2007 at Children's Hospital of Philadelphia. Dr. Bennett is the Scientific Director of that study.



Director of the Center for Cellular and Molecular Therapeutics (CCMT)

Katherine High, MD

Dr. High is a world renowned hematologist, a Howard Hughes Medical Institute investigator specializing in blood coagulation, and the Director of the Center for Cellular and Molecular Therapeutics at the Children's Hospital of Philadelphia. Her research focuses on gene therapy approaches to treat rare genetic diseases, in particular hemophilia. She is a leading researcher for "bench to bed" studies. Her foresight and experience in translational research is unique and irreplaceable.

Director of the Ophthalmic Evaluation and the Genetic Diagnostic Services

Eric A. Pierce, MD, PhD

Dr. Eric Pierce is a fellowship-trained pediatric ophthalmologist with 11 years of practice experience at the Children's Hospitals of Boston and Philadelphia. He is the Director of the CHOP-Penn clinical center for pediatric inherited retinal degenerations. He is also a molecular geneticist and has extensive experience studying photoreceptor biology and degenerations. He will also work with the genetic counselor in Module 3 to obtain accurate molecular diagnosis and to provide counseling to the patients and families and he will perform the clinical examinations of children seen at the Eye on Jacob (Module 1). He will also supervise the collection of electrophysiology data, and coordinate assessments of visual function and retinal structure of patients.

Director of the Communication Disorders Center

Carol Knightly, Au.D. CC-A

Carol Knightly is the Director of Clinical Operations at the Center for Childhood Communications at The Children's Hospital of Philadelphia. She is certified to carry out speech, language and hearing evaluations and has been involved with development of the newborn hearing screening program in the USA.

ADDITIONAL PERSONNEL (CHOP/UPENN)

OPHTHALMIC EVALUATION SERVICES

Albert Maguire, MD

Dr. Albert Maguire is a Vitreo-Retinal Surgeon and is a senior member of the Retina Service at both the Scheie Eye Institute at University of Pennsylvania and in the Department of Pediatric Ophthalmology at the Children's Hospital of Philadelphia (CHOP). He is also a senior investigator at the F.M. Kirby Center for Molecular Ophthalmology. Dr. Maguire has had a long-standing interest in retinal gene transfer/gene therapy and developed surgical approaches with which to deliver genes in proof-of-concept studies involving gene therapy. He has trained numerous investigators at dozens of institutions nationally and internationally on technical aspects of these procedures and has extensive experience performing subretinal injections of viral vectors. Dr. Maguire has published widely on applications of gene transfer approaches for retinal degenerative and ocular neovascular diseases. Dr. Maguire has participated in numerous clinical trials for retinal diseases. His research, conducted at Penn over the past 16 years, has resulted in approval and implementation of gene therapy clinical trials in the United States for Leber Congenital Amaurosis. He is currently the Principal Investigator of a Phase 1 gene therapy clinical trial for this disease.

Kathleen Marshall (Clinical Coordinator)

Kathleen Marshall has worked closely with the clinical staff of Pediatric Ophthalmology at CHOP and with the investigators developing gene therapy treatment for LCA-RPE65 at the Center for Cell and Molecular Therapy (CCMT) at CHOP. She thus serves as an important intermediary between bench researchers, vector development team, pediatric ophthalmologists, ophthalmic technicians and the clinical trials office in this program. She has had in-depth, first hand and formal training in ICH Good Clinical Practice to ensure the welfare of research participants and integrity of the data collected. Mrs. Marshall will assist the Clinical Coordinator in developing the Eye on Jacob Program.

Marianne Letterio, RN, BSN, CCRC (Clinical Coordinator for Eye on Jacob Initiative)

Marianne Letterio has experience coordinating pediatric ophthalmology clinical trials, having served as the clinical coordinator of several retinopathy of prematurity studies at CHOP. She also has laboratory experience and experience in managing regulatory documents for the CHOP clinical trials program. She will work closely with Kathleen Marshall to jumpstart a program for Usher Syndrome. Ms. Letterio will be the liaison between the clinical and research staff and the patients/families. She will coordinate the visits of patients/families with Usher Syndrome and interface with the activities of the other Modules. This will include managing the clinical database (Module 1), managing the scheduling of the patient visits and tests, and, developing information packets and a web page for families, and planning the annual Eye on Jacob Family meeting.

Sonia Zhu, MD

Sonia Zhu is a certified ophthalmic technologist who has been on the clinical staff of Pediatric Ophthalmology at The Children's Hospital of Philadelphia for more than 10 years. She was trained as an ophthalmologist and has significant ophthalmology research experience as well. She is proficient at all aspects of visual function testing in infants and children including electroretinography and acuity, color, contrast and visual field testing. In addition, she maintains databases of all test results, images, surgical cases and references for members of the department. Her duties for the CHOP/Penn Pediatric Center for Retinal Degenerations will include testing of visual function for all patients. Dr. Zhu's expertise will be used for electroretinography and visual function assessments in Module 1.

COMMUNICATIONS DISORDERS CENTER

Joy Peterson, Au.D., CCC-A

Joy Peterson is the Manager of Audiology at the Center for Childhood Communications at The Children's Hospital of Philadelphia. She is certified to carry out speech language and hearing evaluations and to evaluate and dispense hearing aids.

GENETIC DIAGNOSTIC CLINIC

Emily Place, MS

Emily Place is a genetic counselor in the Division of Clinical Genetics at CHOP. She has an MS in Genetic Counseling, and runs the ophthalmic genetics clinic at CHOP. As part of her responsibilities, she will work with Dr. Pierce and the Genetics faculty to coordinate genetic evaluations and testing of children with retinal degenerations.

TRANSLATIONAL RESEARCH TEAM

Jeffrey Bedrosian, MD

Jeffrey Bedrosian received his MD from University of Pennsylvania in 2006, where he initiated a project on gene therapy for USH1C in the Bennett Lab. Dr. Bedrosian developed methods for delivering transgenes to the mouse cochlea and is currently testing for efficacy of gene therapy for hearing loss in an animal model of Ush1c. Dr. Bedrosian is currently a resident in otolaryngology.

Jeannette L. Bennicelli, PhD

Jeannette L. Bennicelli is an expert in the design and construction of DNA vectors as well as assays for cellular and protein functions. Dr. Bennicelli works in the Bennett lab to design and construct DNA vectors to be used in translational research seeking to find treatments for blinding diseases of the retina and in deafness in forms of Usher Syndrome. She was integral in the design, construction, and testing of the adenoassociated viral vector that is currently being tested in a Phase I Safety Study in Subjects with Leber Congenital Amaurosis (LCA) at Children's Hospital of Philadelphia and has also designed and validated assays used to monitor biodistribution and potential immunologic responses to the vector.

Daniel C Chung, DO, MA

Daniel Chung is a trained pediatric ophthalmologist who has concentrated his efforts in gene therapy research for retinal diseases. He has expertise in retinal and cochlea surgery in animal models for development of proof-of-concept for efficacy of gene therapy. Dr. Chung is also a member of the team conducting a gene therapy clinical trial for Lebers Congenital Amaurosis. Dr. Chung is currently in collaborations on development of retinal gene transfer approaches with the University of Iowa, Stanford University, The Jackson Laboratories and with other departments at the University of Pennsylvania.

Michael Anne Gratton, PhD

Michael Anne Gratton began her career as a Clinical Audiologist, but subsequently trained in the basic science and physiology of hearing. She is currently in the Otolaryngology Head-Neck Surgery Department studying the relationship of basement membrane to inner ear function and hearing. Her expertise in cochlear anatomy and physiology has led to collaborations nationally in which her lab assesses auditory function followed by histological analysis of the inner ear in rodent models of hearing loss. She is part of a team evaluating efficacy of gene therapy for hearing loss in animal models of Usher Syndrome.

Arkady Lyubarsky, PhD

Arkady Lyubarsky is a Senior Research Investigator in the Department of Ophthalmology at the University of Pennsylvania School of Medicine. Before joining the University of Pennsylvania research team, he had spent about 20 years with the Academy of Sciences of the USSR where he was performing research on biophysics and physiology of vision. Dr. Lyubarsky has had a long-standing interest in connecting basic research with therapeutic developments. He is currently working on the design and implementation of methods for evaluation of efficacy of gene therapy in protecting and/or restoring the visual function.

Anthony DeLong (Project Administrator)

The coordination of activities between the various modules, Centers and institutions will require a full-time administrator. Anthony DeLong is a business administrator at University of Pennsylvania and is familiar with issues relating to both laboratory research and clinical trials. Mr. DeLong will also assist with website design and response to patient/family queries.

Zhangyong Wei

Zhangyong Wei is a laboratory technician who will manage the animal colony of Ush1c^{-/-} mice and assist in gene therapy experiments. Ms. Wei has 10 years experience in handling and breeding of these animals, with the assays that will be used to measure restoration of visual and auditory function, and with tissue processing techniques that will be used to confirm expression of the appropriate gene in gene therapy experiments.

CENTER FOR CELLULAR AND MOLECULAR THERAPEUTICS

Federico Mingozzi, PhD, MBA

Dr. Federico Mingozzi is the Technical Director of the Translational Research Core of the Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia. He is responsible for non-clinical studies on the safety and efficacy of virus-mediated gene transfer in small and large animal models of diseases. His studies on immune responses to the transgene product and to the viral vector capsid in experimental animals and human subjects constitute fundamental work for the field of AAV-mediated gene therapy.

Junwei Sun, MS, MBA

Mr. Sun is the Administrative Director of the Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia. He is responsible for the 6 logistic and financial planning of the Center's clinical studies. His creative and "out-of-box" thinking has been instrumental to the smooth operation of the translational studies.

Jennifer McDonnell-Wellman, MS

Ms. McDonnell-Wellman is the Associate Director of Regulatory Affairs for the Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia. She has years of experience working with regulatory organizations at both the federal level, including FDA, OHRP, and NIH/OBA, and local level, such as IRB/IEC's, CTTC's, and IBC's. Her efficiency and organizational skills are pivotal at keeping trial timelines on schedule. She is an integral part of many of the on-going, multi-site gene therapy clinical trials sponsored by CCMT.

Fraser Wright, PhD

Dr. Wright is the Director of the Clinical Vector Core and Research Associate Professor of Pathology at the University of Pennsylvania. He has been involved in the development and production of biologics, including recombinant adeno-associated viral (AAV) vectors, avian pox viral vectors, respiratory syncytial viral vectors, and plasmid DNA for the treatment or prevention of serious human diseases for more than ten years. He has led the design and establishment of a BSL2 laboratory suite to produce recombinant viral vectors to support human clinical studies.

Shangzhen Zhou, MD

Dr. Zhou is the Technical Director of the Research Vector Core of the Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia. She is responsible for manufacturing many varieties of AAV viral vector with a high throughput time. Her AAV viral vectors have been widely used in the gene therapy field, and have one of the highest standards in purity and yield.

PATIENT NETWORK TEAM

Jackie Brennan

Jackie Brennan is an Educational Consultant for Hand in Hand, a Pennsylvania Training and Technical Assistance Network. Ms. Brennan also runs the "Little Rock Foundation" family resource room at The Children's Hospital of Philadelphia. This resource is dedicated to supporting families of children with disabilities.

EYE ON JACOB PROGRAM COLLABORATORS AT OTHER INSTITUTIONS

Jose Sahel, MD

Dr. Sahel is the Chairman of Ophthalmology, Director of Clinical Investigation Center, Quinze-Vingts National Ophthalmology Hospital, Director National Reference Center for Retinal Dystrophies; Head of INSERM, University Hospital, Paris, France; Director of the European Union initiative to develop treatments for Usher Syndrome.

Edwin M. Stone, MD

Dr. Stone is a Medical Retina and Inherited Retinal Degeneration Specialist; Director of the John and Marcia Carver Nonprofit Genetic Testing Laboratory; Howard Hughes Medical Investigator, University of Iowa.

Christine Petit, MD

Dr. Petit is the Co-Director of the European Union initiative to develop treatments for Usher Syndrome 7; Unité de Génétique et Physiologie de l'Audition INSERM, Institut Pasteur, Paris, France.





Department of Ophthalmology
Scheie Eye Institute

Jean Bennett, M.D., Ph.D.
Professor of Ophthalmology
Vice Chair for Basic Research

F.M. Kirby Center for Molecular Ophthalmology

December 2008

Dear Friends of "Eye on Jacob":

With the remarkable progress over the past decade in deciphering the genetic bases of different forms of Usher's Syndrome and the development of technology allowing efficient and stable gene transfer to the retina and the cochlea, the time is ripe to develop new treatment strategies for the blindness and deafness that is the hallmark of this disease. Our entire team of physicians, scientists and clinical trial experts is excited about the imminent establishment of the **Eye on Jacob Funds for Usher's Syndrome at UPENN and CHOP**, a comprehensive clinical treatment, research and resource center for individuals with Usher's Syndrome. With the multidisciplinary and complementary expertise and experience of the members of this team, we feel confident that it will be possible to move forward quickly to provide state-of-the-art clinical care and support to families facing Usher's Syndrome. Simultaneously, it will be possible to carry out the studies that will lead ultimately to treatments for this set of diseases for which there are currently limited options.

This is a very exciting and historic time! I predict that the services and research established through the Eye on Jacob Funds will become the model for other groups aiming to develop innovative treatments for inherited conditions for which there currently are none.

Sincerely,

A handwritten signature in blue ink that reads "Jean Bennett". The signature is fluid and cursive, with a long horizontal stroke extending to the right.

Jean Bennett, MD, PhD



Myrin Circle • 51 North 39th Street • Philadelphia, PA 19104-2689 • Tel: 215-898-0915 • FAX: 215-573-7155 • jebennet@mail.med.upenn.edu
On the University of Pennsylvania Presbyterian Medical Center Campus



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 Funding Research for Usher Syndrome

RESEARCH FUNDING OPPORTUNITIES

A one month study for a specific form of Usher Syndrome	\$ 10,000
Genetic testing on 10 individuals with early onset (Type 1) Usher Syndrome	\$ 25,000
Designing the prototype (research-grade) medicine that can be used to rescue vision/hearing loss in Usher Syndrome	\$ 50,000
Multi-focal electroretinogram (mfERG) equipment that will be used to quantify the improvement in vision after gene transfer	\$ 100,000
Determining that the gene therapy approach that will be used in a human clinical trial will be as safe as possible	\$ 250,000
Generating the medicine which could reverse blindness/deafness in one form of Usher Syndrome	\$ 500,000
A human clinical trial for one form of Usher Syndrome including obtaining approvals for the study, performing clinical evaluations, procedures and laboratory tests, personnel, patient travel and lodging costs of the many participants	\$1,000,000



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EYE ON JACOB

Funding Research for Usher Syndrome

CONTRIBUTIONS CAN BE MADE PAYABLE TO:

Eye on Jacob
P.O. Box 162
La Canada, CA 91012

FOR MORE INFORMATION, PLEASE CONTACT:

Mrs. Sonia Desormeaux
P.O. Box 162
La Canada, CA 91012
TEL: 818.669.3949
FAX: 818.952.3932



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