

## **FAQ**

### **How is the proposed study different from a microarray (chromosomal microarray analysis, CMA)?**

A microarray can identify large, deleted or added regions of chromosomes but cannot precisely determine the specific sets of genes that are missing, nor detect pieces of other chromosomes that can sometimes take the place of the deleted region. For this project, we propose to use a personalized medicine strategy that takes advantage of advanced DNA sequencing technologies as well as cellular-based experiments to help understand precisely which genes are most critical in defining specific characteristics of individuals with chromosome 9 P Minus Syndrome. Your child's whole genome will be sequenced.

### **Can we participate if we're not in the US?**

Unfortunately, at this time this is not an option as shipping blood internationally is not easily done. If you plan to be at the 9p- event in Iowa next July, then you could submit a blood sample then.

### **How is the study being funded?**

The first part of the study which encompasses the collection of samples from the 9p- population and the initial analysis of key genes is funded by an anonymous donor. Subsequent studies which will look at further isolation of specific genes to determine function will require additional funding that will hopefully be obtained by interested investigators who will apply for competitive research support.

### **We gave saliva 2 years ago at the summer meeting in Louisiana, do I need submit a sample again?**

Yes, because the sample donated then was saliva, and this new study will require blood, given the more robust tests entailed.

### **Which doctors are responsible for running the study?**

Drs. Jeff Milbrandt and Rob Mitra from the Washington University Department of Genetics will be running the analysis. They are working closely with Dr. Francis Sessions Cole who has worked with the 9p minus group for a long time.

### **What exactly will you need from us to participate?**

To participate in this research project, each individual (or her/his parent or legal guardian) will need to provide informed consent to become a research subject. They will also need to provide a detailed summary of her or his medical and family histories and specific characteristics through a standardized examination and/or questionnaire. You will then send in a blood sample drawn from the arm. We will provide a sample kit and prepaid airbill so you can send us the results.

### **How will I be kept up to speed on progress?**

We plan to use the 9pminus research Facebook group to keep individuals abreast of the research. Participants will also receive information directly once the results of their samples are ready. We understand that it is dispiriting to families to go through the process of donating a sample and then not hear back about research progress.

### **Is this a cure for 9p- syndrome?**

No, there is no "cure," and importantly, it's unlikely that one will emerge in the short term. Through these studies conducted over the next several years, we plan to identify the most important missing genes as they will serve as high priority targets in development of future treatment strategies for people with chromosome 9 P Minus Syndrome

**How long will it take before we get any tangible results?**

We hope that within 3 years from the start of the research project, we will have some tangible results.

**Will I be given an individualized result for my child, i.e., a report showing exactly which genes are affected?**

Participants will be provided with updates on the laboratory research being performed related to their condition. Because this project is a research project, individual test results cannot be used for clinical decision making.

**How many samples do we need to collect to make the study meaningful?**

Our goal is to get to 100 samples which should make for a sufficiently robust data set to draw initial conclusions on the importance of missing genes. A smaller number will still work, but let's try and get that 100!

**How will the blood samples be collected?**

Once you have agreed to participate, you will be mailed a kit with a pre-paid shipping label and a test tube. You can then either have your pediatrician draw the blood, and you can send it to Washington University. Alternatively, we have arranged with OneHealth (a division of Quest Laboratories) to come draw the sample at a time of your choosing at your home. They will then be responsible for sending the sample to the lab at Washington University.

**I am a doctor or scientist and think I can help contribute to the research project; how can I help?**

Please contact Dr. Cole directly ([fcole@wustl.edu](mailto:fcole@wustl.edu) or 314-454-6183) if you would like to contribute.

**Are there any drawbacks from participating?**

Other than going through the trouble of having the blood drawn and sending in the blood sample, there's little other impact to participants. Their samples will help us better understand the impact of each missing gene, and in the long run hopefully this contribution pays off through more personalized medicine options.

**Will a participant's data be made public or available to others?**

Data will be anonymized and kept confidential. No identifiable individual data will be published.

**I would like to contribute to the funding of the study, how can I do so?**

Thankfully, we have the support of the 9p- group, and they've conveniently set up a charity to allow us to fund this work. Please contact Ciera Dickson ( [cdickson@9pminus.org](mailto:cdickson@9pminus.org) ) or Board members of the Chromosome 9 P Minus Network for additional details on how this can be done.

**Can I withdraw from the study if I change my mind?**

You're free to withdraw from the project without giving a reason. If you choose to withdraw, you or your child will still hear about any progress that we make, but the specific information about your child will not be used.

**Ok, you've convinced me. Now what?**

Please email Tori Tate ( [victoriatate@wustl.edu](mailto:victoriatate@wustl.edu) ) to note your interest in participating. Please also note if you would be willing to cover the costs of the sample collection kit and shipping [we might provide an estimated cost?]. Once we're ready to go, we'll contact you!

**Doctor bios:****Francis Sessions Cole III, MD**

F. Sessions Cole, III, MD, is a neonatologist who is the Park J. White, MD, Professor of Pediatrics, Assistant Vice Chancellor for Children's Health, Executive Vice Chairman of the Department of Pediatrics, and Interim Chief of the Division of Allergy, Immunology, and Pulmonary Medicine at Washington University School of Medicine.

Dr. Cole is also known for his remarkable dedication as a clinician, researcher and educator. He has been recognized by his peers in *Best Doctors in America* and *America's Top Doctors*, chosen as the recipient of numerous teaching awards by his medical students and residents, and is a National Institutes of Health (NIH)-funded investigator whose research has provided a better understanding of the genetic basis of rare, inherited diseases. For example, his findings have made him one of the world's leading authorities on the genetic causes of respiratory distress in newborns.

Dr. Cole received his bachelor's degree from Amherst College in 1969 and graduated from Yale Medical School in 1973. He completed his internship and residency at Boston Children's Hospital/Harvard Medical School and served as a General Medical Officer in the United States Public Health Service.

He was a fellow in neonatology at The Joint Program in Neonatology/Harvard Medical School (Boston Children's Hospital, Brigham and Women's Hospital, and Beth Israel Hospital) before becoming Assistant Professor of Pediatrics at Harvard Medical School in 1982. In 1986, he joined the Washington University School of Medicine as Associate Professor and Director of the Division of Newborn Medicine. He was promoted to professor in 1989 and named vice chairman of the department in 1995.

**Jeffrey Milbrandt, MD, PhD**

Jeffrey Milbrandt, MD, PhD, the James S. McDonnell Professor of Genetics and head of the Department of Genetics, leads faculty members widely recognized for their work to uncover fundamental genetic mechanisms that underlie biological processes and disease. Milbrandt, who applies genetic tools and genomic technologies to his own studies, has devoted his career to Washington University. His research focuses on neuronal signaling cascades in an effort to understand how specialized nerve cells called glial cells contribute to nerve regeneration after injury. In a longstanding collaboration with Eugene Johnson Jr., PhD, professor of neurology, Milbrandt's laboratory discovered a family of growth factors that promote neuronal survival. One member of this family, neurturin, is being evaluated in clinical trials in patients with Parkinson's disease. Dr. Milbrandt's studies have also pointed to the vulnerability of axons, which transmit signals between nerve cells, in neurodegenerative diseases such as Parkinson's, amyotrophic lateral sclerosis (ALS) and Alzheimer's. Recently, he has discovered several molecules that can protect axons from degenerating after an injury. These include enzymes, a protein thought to extend life called SIRT1 and resveratrol, a minor ingredient in red wine. He is now working with biotech companies to test therapies that combine growth factors with axonal

protective agents as a potential treatment for neurodegenerative diseases.

Dr. Milbrandt was also actively involved in the establishment of the Hope Center for Neurological Disorders, a collaboration that pools the intellectual and financial resources of Washington University and Hope Happens for Neurological Disorders. The center is dedicated to supporting and accelerating research that uncovers molecular mechanisms that contribute to neurodegenerative diseases and uses these discoveries to develop new treatments for diseases.

### **Robi Mitra, PhD**

Rob Mitra, PhD, is the Alvin Goldfarb Professor of Computational Biology in the Department of Genetics and the Edison Family Center for Genome Sciences and Systems Biology at Washington University in St. Louis. He received his BS, MS and PhD from MIT. His current research interests are focused on understanding how transcription factors achieve their in vivo specificities, developing new genomic technologies, and applying these to understand disease processes.

### **Tychele Turner, PhD**

Dr. Tychele Turner is a geneticist/genomicist with a deep interest in understanding the genetic architecture of human disease. Dr. Turner completed her undergraduate training in Genomics and Molecular Genetics at Michigan State University followed by graduate work at the Johns Hopkins University School of Medicine where she received her Ph.D. in Human Genetics and Molecular Biology under the mentorship of Dr. Aravinda Chakravarti. During graduate work, Dr. Turner focused on the genetics of autism in females. Dr. Turner performed her postdoctoral work in the laboratory of Dr. Evan Eichler in the Department of Genome Sciences at the University of Washington. There she performed large-scale whole-exome and whole-genome sequencing analyses to discover risk factors for autism. Currently, Dr. Turner is an Assistant Professor in the Department of Genetics at Washington University School of Medicine in St. Louis, Missouri. She is actively involved in the 9p project and is focused on breakpoint resolution and genotype/phenotype correlations.

### **Patricia Dickson, MD**

Patricia Dickson, MD, is the Centennial Professor of Pediatrics at Washington University School of Medicine. She also is professor of genetics and chief of the Division of Genetics and Genomic Medicine in the Department of Pediatrics. She earned an undergraduate degree from the University of Chicago and her medical degree from Columbia University College of Physicians and Surgeons. She completed an internship and residency at Harbor-UCLA Medical Center, a Los Angeles County hospital affiliated with UCLA and the Los Angeles Biomedical Research Institute. She served as chief resident at the medical center and completed her medical genetics training in the UCLA Intercampus Medical Genetics Training Program. Dr. Dickson's research focuses on pathogenesis and novel therapy development for central nervous system disease due to the genetic disease mucopolysaccharidosis. She has been successful in bringing some of these therapies to clinical trials. To date, Dr. Dickson's work has led to 60 publications and has been cited more than 1,000 times. Her honors include the National Institutes of Health National Research Service Award Fellowship and the Richard B. Weitzman Memorial Award for

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Meritorious Research. She is a diplomate of the American Board of Pediatrics and the American Board of Medical Genetics and Genomics.