NEURAL TUBE DEFECTS (NTD) are among the most common birth defects and affect approximately 1-2 infants per 1,000 births in the United States. An NTD is an opening in the spinal cord or brain that can often be surgically closed at birth. However, patients with NTD often have permanent nerve damage and disability. An NTD can develop before a woman may even know she is pregnant, and in women from all racial, ethnic, and social groups.

Research has shown that 50-70% of NTD can be prevented when women supplement their diet with folic acid, a water-soluble B-Vitamin. For that reason, most research has focused on genes in the folate-metabolism pathway. However, we still understand very little about which genes are involved and how they function. Researchers also believe that genes in other pathways may play equally important roles. HIHG researchers are searching for these genes and trying to understand how the environment contributes to development of NTD.

We hope that our research will allow us to better understand the genetic and environmental causes of NTD, which will eventually lead to more accurate genetic counseling and risk assessment, improved treatments, better prevention methods, and hopefully a cure.
WHAT IS OUR RESEARCH GOAL?
HIHG researchers are trying to identify the genetic and environmental causes of Neural Tube Defects (NTD). We are interested in learning more about the causes of both open NTD like spina bifida, anencephaly, and encephalocele, and closed NTD like lipomeningocele, lipomyelomeningocele, and tethered cord.

HOW CAN YOU HELP?
In order to identify these genetic and environmental factors we require the participation of a large number of individuals. If you, a family member, or friend has been diagnosed with an NTD such as spina bifida, we invite you to join our research studies.

WHO CAN PARTICIPATE?
• Individuals diagnosed with NTD
• Parents or siblings of the individual(s) with NTD
• Friends or caregivers who have not been diagnosed with NTD to serve as healthy controls

WHAT DOES PARTICIPATION INVOLVE?
Participants will be asked to:
• Read and sign a consent form
• Provide a blood sample
• Participate in a family and medical history interview
• Grant research staff permission to review the medical records of the individual(s) with NTD

DO FAMILIES GET STUDY RESULTS?
Although we are unable to provide individual results, you will receive newsletters that describe our research progress and discoveries related to NTD.

STUDY PARTICIPATION FACTS
• Participation is voluntary
• All information is kept strictly confidential
• There is no cost to participate
• Travel to the University of Miami Miller School of Medicine is optional, but not required
• Joining the study will not affect your healthcare or insurance

THE RESEARCH TEAM
To make this research project a success, we need many people with different areas of expertise. Individuals, families, physicians, genetic counselors, and human geneticists are all working together to find the genes that cause NTD.

Our team is led by co-principal investigators Evadnie Rampersaud, Ph.D., a genetic epidemiologist, and John Gilbert, Ph.D., a molecular geneticist, along with Margaret A. Pericak-Vance, Ph.D., Director of the HIHG. Members of the HIHG lead the field in identifying the genetic variants that underlie common human disease. They have identified genes for more than 60 human illnesses, including neurological disorders, cardiovascular diseases, and eye diseases.

CALL OUR TOLL-FREE NUMBER
877-686-6444