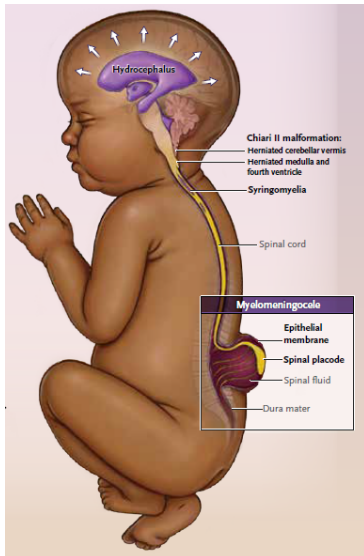


Why are Researchers doing this Study?

Spina bifida is a congenital disorder with significant long-term complications and is the second most common congenital anomaly.

We know that spina bifida is inherited, but a consistent genetic cause has not been identified. In addition, we know some environmental factors can cause spina bifida, including drugs (*i.e.*, medicines), maternal diabetes, and exposure to heat. This information suggests that the inheritance of spina bifida might be epigenetic through a process called DNA methylation, which can be both affected by environmental factors and inherited. The purpose of this research is to study whether spina bifida is inherited via DNA methylation.



Department of Neurological Surgery

UNIVERSITY OF WISCONSIN
SCHOOL OF MEDICINE AND PUBLIC HEALTH

**Contact us for more information
on this study:**

Sima Sayyahmelli, MD

Researcher I

Office: 600 Highland Ave, K4/848 CSC

Madison, WI 53792-8660

Phone: (608) 265-4514

Email: sayyahmelli@neurosurgery.wisc.edu



<https://go.wisc.edu/2vf4ww>

*Disclaimer: Email is not a secure form of communication. Please avoid sending detailed sensitive information via email.



Department of Neurological Surgery

UNIVERSITY OF WISCONSIN
SCHOOL OF MEDICINE AND PUBLIC HEALTH

DNA METHYLATION IN SPINA BIFIDA

Study

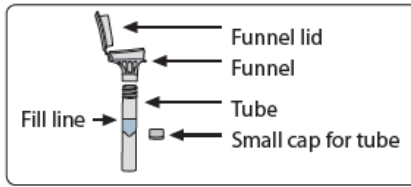
Protocol Number: 2022-1027

Principal Investigators (PIs):

Bermans Iskandar, MD

Reid Alisch, PhD

Funding for this study is provided by ICTR (UW Institute for Clinical and Translational Research)



Medical records review We will also collect information from your medical records such as your family and medical history, and consult with the researcher involved in the study, to make sure no one in your family has the disease. This information will not be shared with anyone outside of the study team, and the publications that result from the study will not divulge personal information about you such as your name.

Benefits

Joining this study will help us better understand the cause of spina bifida. Potential benefits to society include a better understanding of this disease from a genetic point of view, which would help with prevention and genetic counseling.

Invitation to Participate in a Research Study

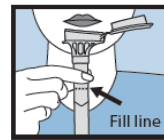
We invite you to take part in a research study about spina bifida. Participation is voluntary and participants can withdraw from the study at any time. You can participate as a patient with spina bifida, or as a control volunteer (without spina bifida).

Study Methods

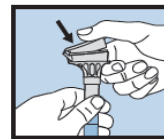
Participants: Spina bifida patients and controls between the ages 15 and 45 will be included in this study.

Specimen collection: If you agree to participate in this research study, the researchers will ask you to provide approximately 1-2 milliliters saliva for testing.

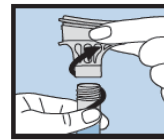
These samples can be completed in clinic, or at home after which the kits will be mailed back to us. A video will be provided that explains how to obtain the saliva.



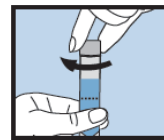
1 Spit into funnel until the amount of liquid saliva (not bubbles) reaches the fill line shown in picture #1.



2 Hold the tube upright with one hand. Close the funnel lid with the other hand (as shown) by firmly pushing the lid until you hear a loud click. The liquid in the lid will be released into the tube to mix with the saliva. Make sure that the lid is closed tightly.



3 Hold the tube upright. Unscrew the funnel from the tube.



4 Use the small cap to close the tube tightly.



5 Shake the capped tube for 5 seconds. Discard or recycle the funnel.

Study Hypothesis

Environmental factors can influence and contribute to heritable diseases without a change in the gene.

These observations suggest that environmental modifications may be a cause of spina bifida and its inheritance. In this study, we want to examine this possibility.

Participants

Individuals with spina bifida will be recruited from the clinical population at University of Wisconsin. We will collect the information from your medical records such as your family and medical history for pre-screening eligibility.

Volunteers/Controls You don't have the disease, but obviously we need normal subjects who would be used as controls. Controls will be recruited from age- and sex-matched friends of spina bifida patients. Each spina bifida participant will be asked to invite a friend who is age and sex matched to consider participating as a control. If they agree, the patient will ask them if contact information can be shared with the study team.

