WHOLE GENOME AND RNA SEQUENCE ANALYSIS IN PRADER-WILLI SYNDROME

The Foundation for Prader-Willi Research (FPWR) is conducting a research study to better understand how variations across the entire genome influence the range and severity of symptoms in people with Prader-Willi syndrome (PWS).

WHAT ARE THE GOALS OF THE STUDY?

- The clinical and behavioral features of PWS occur with varying frequencies and severity between affected individuals.
- The goal of this study is to identify genetic variants that influence the risk and severity of these symptoms in individuals with PWS.
- The information from this study may eventually be helpful for PWS families, clinicians, and the scientific community to better understand individual risk for PWS symptoms, as well as response to treatments.

WHO CAN PARTICIPATE?

Participants must:

- Have a genetically confirmed diagnosis of PWS
- Be age 10-65 years
- Reside in the United States
- Be enrolled in the Global PWS Registry and have all surveys completed or updated within the past 3 months

WHAT IS INVOLVED?

- Review study information and consent to the study
- Complete (or update) all surveys in the Global PWS Registry and upload a confirmed PWS diagnosis
- Provide copies of medical records if requested
- Provide a blood sample from the person with PWS (either at home or at the doctor). This may be blood spots on a piece of filter paper, or in some cases, a blood draw (~1/2 to 2 teaspoons).
- Participants will receive a brief summary of individual results in 9-12 months after the blood sample. We will discuss these results with you by telephone or video conference.

If you are interested in participating in this study, please contact Caroline Vrana-Diaz (caroline@fpwr.org).

Approved
April 21, 2021
North Star Review Board