

(MRI) of the brain and cervical (neck) spinal cord to measure the size of the head and determine the presence of the Chiari I malformation and syringomyelia. A small blood sample (about 2 tablespoons) will be drawn for DNA studies relating to the Chiari I malformation.

Type I Arnold Chiari Malformation

Official Title: Genetic Analysis of the Chiari I Malformation

Study Start Date: February 2000

Objectives: The goal of this study is to establish family pedigrees and undertake genetic linkage analysis that will identify gene loci associated with the Chiari I malformation and underdevelopment of the bone forming the posterior cranial fossa.

Study Population: Patients and family members of patients with the Chiari I malformation. Because the research institutions are located in the United States and Russia, subjects will be recruited predominantly from these countries.

Design: Human subjects will undergo 1) neurologic examinations, 2) head and cervical MRI scans to evaluate for the Chiari I malformation, syringomyelia, and maldevelopment of the posterior fossa, and 3) isolation and analysis of genomic DNA from whole blood for linkage analysis.

Outcome Measures: Pedigrees will be established based on