

SMA Research Update – Providing Hope!

Spinal Muscular Atrophy (SMA) Biomarkers in the Immediate Postnatal Period of Development

Principal Investigator, Claudia Chiriboga, MD, MPH

COMPLETED

Spinal muscular atrophy (SMA) is the leading genetic cause of death in infants. Preclinical evidence strongly suggests that effective therapy must be delivered as early as possible to prevent disease progression. The Columbia University SMA Clinical Research Center is partnering with the [NeuroNEXT](#) clinical trial network to conduct this study. The primary objective of this study is to identify [biomarkers](#) to better understand the disease and how it manifests and aid researchers to most effectively design and execute SMA clinical trials in infants.

What is a Biomarker?

A Biomarker is any laboratory measurement that reflects the activity or stage of a disease process. For example, cholesterol levels reflect heart disease risk. Biomarkers are being used effectively for cancer, diabetes, and heart disease to help determine whether new drugs are helping patients, and to better understand the diseases. The goal of the SMA Biomarker Project is to identify laboratory measurements that can be used in future SMA clinical trials.

What is involved in this study and how do I know if I qualify?

The study involves collecting information about your child's development, immediately after birth up to 24 months of age. Children enrolled in this study will be required to come to the clinic every 3 months (or a maximum of 7 visits) and will undergo a number of procedures at each visit; these include, vital signs and motor function assessments, clinical exam, electrical impedance myography (EIM), compound motor action potential (CMAP) and blood draws. Infants 6 months of age and younger who have been diagnosed with Spinal Muscular Atrophy as well as, healthy infants are encouraged to participate.

To be considered for eligibility, **infants with SMA** must meet the following criteria:

- Birth between 36 and 42 weeks inclusive of gestation
- be between 0-6 months of age at the time of enrollment
- have a documented homozygous SMN1 gene mutation/ deletion. An infant can have any number of SMN2 gene copies
- be able to follow all study procedures
- be healthy
- not require breathing support for over 12 hours
- not have a history of tracheostomy tubes and ventilator-dependency

- Parents or guardians must be able to sign an informed consent prior to any study procedure being performed

Healthy control infants who meet the following criteria will be enrolled:

- Birth between 36 and 42 weeks inclusive of gestation
- Siblings of children with SMA must have had prior SMA genetic testing completed con-firming the infant is a healthy control
- Principal investigator feels the family/infant is able and willing to comply with study procedures
- Parent or guardian able to give informed consent

This study is being conducted by the SMA Clinical Research Center in collaboration with Ohio State University and fifteen other centers nationwide. If you would like more information about this study please contact Nicole Holuba, MSN at nh2282@columbia.edu or **212-304-5205**.