NUCHAL TRANSLUCENCY MEASUREMENT IN FETUSES WITH SPINAL MUSCULAR ATROPHY
Neda Zadeh, Louanne Hudgins, Mary E. Norton

Division of Medical Genetics, Stanford University, Stanford, CA, United States, Perinatal Genetics Services, Kaiser Permanante, San Francisco, CA, United States

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder characterized by progressive degeneration and loss of anterior horn cells in the spinal cord. Prenatal molecular diagnosis of this disorder is routinely performed after a previously affected case has been identified in the family. A short case report by Stiller RJ et al. (1999) described a fetus with SMA with increased nuchal translucency (NT) measurement at 13 weeks gestation and cited five additional cases previously published with NT measurements ranging from <3.5 mm to 9.6 mm in the medical literature. The purpose of our study is to determine whether an increased NT measurement is associated with an increased risk for SMA.

Following IRB approval, we enrolled women who had NT ultrasound during pregnancies known to be affected with SMA. All offspring were confirmed via molecular testing. Participants learned of our study through various SMA family support groups. Twenty-eight mothers of children with SMA contacted us to take part in the study. Preliminary review of our data revealed 11 cases with demonstratable data (i.e. NT results and molecular testing) that met enrollment criteria. All women had normal NT values of less than or equal to 2.4 mm with a mean of 1.8 mm for their affected fetuses. Thus at this time, we have not established correlation of predictive value of NT measurement and SMA in our patient population. As this was a small preliminary study, a larger patient sample is warranted to yield more defined results before recommendations can be made.

References: