

# SMA Research Update – Providing Hope!

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## Currently, there are (7) active studies within Columbia University Medical Center:

### **Phase 3, Open-Label, Gene Replacement Therapy Clinical Trial for Patients With Spinal Muscular Atrophy Type 1 (STR1VE)**

#### **\*\*Active and recruiting\*\***

Phase 3, open-label, single-arm, single-dose, study of AVXS-101 (gene replacement therapy) in patients with spinal muscular atrophy (SMA) Type 1 who meet enrollment criteria and are genetically defined by nonfunctional survival motor neuron 1 gene (SMN1) with 1 or 2 copies of survival motor neuron 2 gene (SMN2). Fifteen (15) patients < 6 months (< 180 days) of age at the time of gene replacement therapy (Day 1) will be enrolled.

#### Eligibility

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**Ages Eligible for Study:** up to 180 Days (Child)  
**Sexes Eligible for Study:** All  
**Accepts Healthy Volunteers:** No

#### **Inclusion Criteria:**

- Patients with SMA Type 1 based on gene mutation analysis with bi-allelic SMN1 mutations (deletion or point mutations) and 1 or 2 copies of SMN2 (inclusive of the known SMN2 gene modifier mutation (c.859G>C))
- Patients must be < 6 months (< 180 days) of age at the time of AVXS-101 infusion
- Patients must have a swallowing evaluation test performed prior to administration of gene replacement therapy

#### **Exclusion Criteria:**

- Tracheostomy or current use or requirement of non-invasive ventilatory support averaging  $\geq 6$  hours daily over the 7 days prior to the screening visit; or  $\geq 6$  hours/day on average during the screening period or requiring ventilatory support while awake over the 7 days prior to screening or at any point during the screening period prior to dosing

- Patients with signs of aspiration/inability to tolerate non-thickened liquids based on a formal swallowing test performed as part of screening. Patients with a gastrostomy tube who pass the swallowing test will be allowed to enroll in the study
- Patients whose weight-for-age is below the third percentile based on World Health Organization (WHO) Child Growth Standards
- Participation in recent SMA treatment clinical study (with the exception of observational cohort studies or non-interventional studies) or receipt of an investigational or commercial compound, product, or therapy administered with the intent to treat SMA (e.g., nusinersen, valproic acid) at any time prior to screening for this study. Oral  $\beta$ -agonists must be discontinued at least 30 days before gene therapy dosing. Inhaled albuterol

Estimated Enrollment: 15

Actual Study Start Date: September 29, 2017

Estimated Study Completion Date: March 31, 2020

Additional participating locations:

Stanford University (California), Children's Hospital Colorado (Colorado), Ann and Robert H Lurie Children's Hospital (Illinois), Johns Hopkins Pediatric Neurology (Maryland), Nationwide Children's Hospital (Ohio), Oregon Health and Science University (Oregon).

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**Currently there are (29) studies actively recruiting and enrolling patients throughout the world outside of Columbia University, for further information go to [clinicaltrials.gov](https://clinicaltrials.gov).**

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