Gene Transfer Clinical Trial for Spinal Muscular Atrophy Type 1

This study is a phase I, single-site, dose escalation study to evaluate the safety and efficacy of gene transfer for Spinal Muscular Atrophy Type 1 (SMA1). Enrollment is planned to begin within the first quarter 2014 at Nationwide Children's Hospital in Columbus, Ohio. Infants between 0 and 9 months of age with SMA1 may be eligible to take part in this first human trial. A total of nine patients will be enrolled to receive a one-time gene transfer infusion. Patients will continue to be monitored at Nationwide Children's Hospital including physical exams and blood tests for two years after gene transfer.

Eligibility

- Age: Nine months of age and younger
- Must be diagnosed with SMA Type 1 as defined by the following features:

Inclusion Criteria:

- Mutations of the SMN1 gene with two copies of SMN2 (no more and no fewer)
- Onset of disease at birth to 6 months of age
- Weakness of muscles and joints demonstrated at time of enrollment

Exclusion Criteria:

- Active viral infection (includes HIV or serology positive for hepatitis B or C)
- Use of invasive ventilatory support (tracheotomy with positive pressure)* or pulse oximetry <95% saturation.
- Current illness that in the opinion of the researcher creates unnecessary risks for gene transfer
- Current use of any of the following drugs: drugs for treatment of myopathy or neuropathy, agents used to treat diabetes mellitus, or ongoing

immunosuppressive therapy or immunosuppressive therapy within 3 months of starting the trial (e.g. corticosteroids, cyclosporine, tacrolimus, methotrexate, cyclophosphamide, intravenous immunoglobulin, rituximab)

- Patients with Anti-AAV9 antibody titers >1:50 as determined by ELISA binding immunoassay.
- Abnormal laboratory values considered clinically significant (GGT > 3XULN, Bilirubin ≥ 3.0 mg/dL , Creatinine ≥ 1.8 mg/dL, Hgb < 8 or > 18 g/Dl; WBC > 15,000 per cmm)
- Participation in recent SMA treatment clinical trial that in the opinion of the researcher creates unnecessary risks for gene transfer.
- Family does not want to disclose patient's study participation with primary care physician and other medical providers
- Patient with signs of aspiration based on a swallowing test and unwilling to use an alternative method to oral feeding

*Patients may be put on non-invasive ventilator support (BiPAP) for less than 16 hours a day at the discretion of their physician or research staff.

If you have an infant with SMA Type 1 between 0 and 9 months of age and might be interested in participating please contact the study coordinator:

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