Gene Therapy for Late Infantile Neuronal Ceroid Lipofuscinoses (also known as CLN2 Disease or Batten Disease)

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**Background of Batten Disease**
- Autosomal recessive, ~400-600 cases worldwide
- Disease onset ages 2-4
- Cognitive impairment, visual failure, seizures, and deteriorating motor development, leading to a vegetative state and death by ages 8-12
- Caused by Mutations in the CLN2 Gene

**Gene Therapy**
- Means of delivering therapeutic proteins to the CNS
- Persistent gene expression possible with adeno-associated virus vectors
- Required vector doses within current technology
- Limited host responses to vector

**Clinical Assessment**
- Motor + language clinical score
- Quantitative MRI

**Overall Trial Design**

**Screening protocol**
- n=39 children have undergone initial screening

**Vector protocols**
- n=8 children treated on NIH protocol
- n=4 children treated on the “parallel protocol”

**Safety evaluation**
- No unanticipated major adverse events

**Targets for Therapy**
- 6 burr holes, 2 injections (different levels) per burr hole, 2 μl/min, 150 μl at each level, 3 hr total infusion time

**Status of the AAVrh.10 Trial**

**Rate of Decline of LINCL Quantitative MRI Analysis of Treated Subjects vs Motor + Language Scores of Untreated Subjects**

**Metabolite Pathway Enrichment Analysis and Impact View for LINCL**

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