

# Trial Protocol

**Title**

Proof of concept/Analysis of the possibility of translation of stop codons in Mucopolysaccharidosis type III or SANFILIPPO disease

**Research basis**

Research carried out on fibroblast isolated from patients carrying one or more mutations inducing a stop codon

**Protocol :**

The protocol is comprised of two steps:

- CELL BIOLOGY
  - 1) Defrosting and recovery of the fibroblast strains from the patients
  - 2) Culture and propagation of the strains
  - 3) Harvesting of the cell material and incubation in the presence of the molecular compounds tested.
  
- BIOCHEMISTRY and MOLECULAR BIOLOGY
  - 4) RNA extraction
  - 5) Tests
  - 6) Measurement of enzyme activity
  - 7) RNA studies

**Trial start date**

01/06/06

**Trial end date**

01/03/07