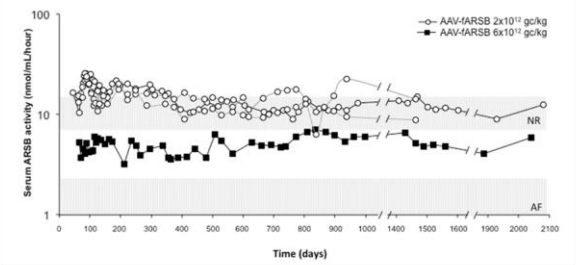
AAV-BASED GENE THERAPY FOR MUCOPOLYSACCHARIDOSIS TYPE VI (MPSVI; MAROTEAUX-LAMY SYNDROME)

This project originates from the Telethon Institute of Genetics and Medicine (TIGEM), **an international reference center for research on genetic diseases** that has established fruitful collaborations with leading companies in the field. The team led by Alberto Auricchio has a **strong track record in the field of gene therapy** and a well-established **network with reference clinicians for the disease**. Prof. Auricchio successfully established and coordinates a network of companies and research centers that was able to secure EU funding under **Framework Program 7** (Grant #304999 MeuSIX, **http://meusix.tigem.it**) for the clinical development of the proposed AAV-based approach to treat MPSVI, an **autosomal recessive** disease caused by mutations in the ARSB gene that affects 1 in 100,000/300,000 live births.



Long-term follow up of serum ARSB activity levels in MPS VI cats treated with AAV2/8.TBG.*felineARSB*. MPS VI cats were injected at postnatal day 50 with various doses of AAV2/8.TBG.*felineARSB*. Each curve represents ARSB values over time from a single animal. The upper and lower grey areas represent the range of values of normal (NR) and affected (AF) cats, respectively.

ADVANTAGES:

- **FIH planned for mid-2016**
- **PoC** reached in **small and large animal models** of the disease
- **Single infusion** leads to **sustained and lifelong effect**, overall treatment **costs are reduced** compared to enzyme replacement therapy
- **Strong connection with the Unit of Pediatrics, Dept. of Translational medicine,** of Università degli Studi di Napoli Federico II where TIGEM PIs have conducted clinical trials for other indications in the past
- Ongoing **GLP Safety and Toxicity** studies (planned to end in Q4 2015).
- **Top record and unique Know-how** in the field of **lysosomal storage disorders**
- **Extensive collection of disease models and cell lines**.

OPPORTUNITY

In line with our mission to develop therapies for patients affected by **genetic diseases**, Telethon is actively seeking **partners** to complement its competencies and bring therapies to the market
We are looking for partners for the **clinical development** of the proposed gene therapy approach
The therapeutic approach has obtained **orphan drug designation** from FDA and EMA

References:

Tessitore et al., Mol Ther. 2008 Jan;**16(1)**:30-7
Cotugno et al., Hum Gene Ther. 2010 May;**21(5)**:555-69
Cotugno et al., Mol Ther 2011: **19(3)**, 461-9
Ferla et al., Hum Gene Ther 2013: **24(2)**, 163-9
Ferla et al., Hum Gene Ther 2014: **25(7)**, 609-18

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