2ND MULTI-STAKEHOLDER Symposium ON IMPROVING PATIENT ACCESS TO RARE DISEASE THERAPIES

22-23 FEBRUARY 2017
HOTEL LE PLAZA
BRUSSELS

20 years EURORDIS
RARE DISEASES EUROPE

Case study: The importance of multi-stakeholder collaboration

Lucia Monaco

Chief Scientific Officer, Fondazione Telethon, Italy

#RareEU2017

Case study: the importance of multi-stakeholder collaboration

2° MULTI-STAKE-HOLDER SYMPOSIUM ON IMPROVING PATIENT ACCESS TO RARE DISEASE THERAPIES
Brussels, 22-23 February 2017

Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy

FONDAZIONE Telethon

May 2016 the first ex-vivo gene therapy treatment approved in the world

FONDAZIONE Telethon

How did we get there?

Strimvelis®▼
(autologous CD34+ cells transduced to express ADA)
for the treatment of patients with ADA-SCID for whom no suitable HLA-matched related stem cell donor is available

ADA-SCID

- Life-threatening disease
- Often fatal within the child's first years of life
- Incidence : 2-7 per million live births

- Severe immunodeficiency caused by mutated ADA gene
- The ADA enzyme protects developing lymphocytes from death

1995: SR-TIGET is founded

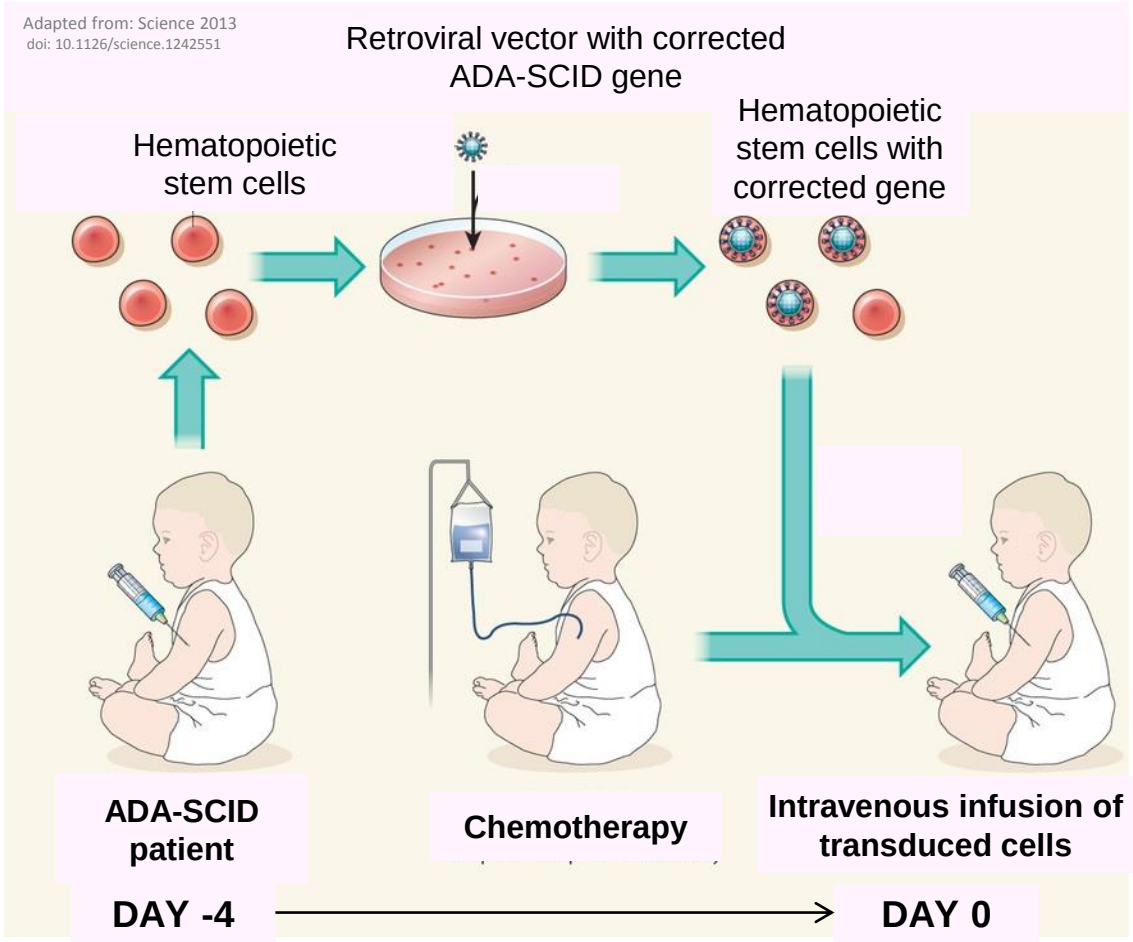
Fondazione Telethon
Italian charity
MISSION: to bring the results of excellent research to patients affected by rare genetic diseases

Ospedale San Raffaele
Major Italian research hospital, Milan

San Raffaele-Telethon Institute for Gene Therapy

- Excellent research
- Innovative gene therapy approaches
- Research AND clinical setting

2000-2009: ex vivo gene therapy clinical trials for ADA-SCID



- **2000**: first patient treated
- **2002-2009**: Pivotal phase I/II clinical trial (12 patients)

- **2000**: GMP vector production at CMO MolMed
- **2005**: EMA ODD
- **2007**: Protocol Assistance from EMA
- **2009**: FDA ODD

ADA-SCID gene therapy is safe and effective

- GT is effective as single therapy long-term
- Decreased rate of severe infections
- Improved T cell counts
- No cases of genotoxicity

How to treat the next patients?

The reasons for an alliance

- **One gene therapy clinical trial (ADA-SCID) successfully completed**
  - Need to **develop production** of medicinal product suitable for **market registration**
- **Six more gene therapy studies at advanced preclinical level (lentiviral platform)**
  - Need to **finalize preclinical studies** and **complete clinical trials** before applying for market registration

- **Financial resources**
- **Competences** and **facilities** for **medicinal product development and production**
- **Marketing authorization**
- **Distribution** and **post-marketing surveillance**

The power of collaboration

1995
SR-TIGET is founded

2010
Alliance with GSK

PARTNERSHIPS

Strimvelis®
(autologous CD34+ cells transduced to express ADA)
for the treatment of patients with ADA-SCID for whom no suitable HLA-matched related stem cell donor is available

ADA-SCID

1995

2000
1st ADA-SCID patient treated
GMP production at CMO MolMed

2002-2009

2002-2009
Phase I/II trial

2010

2010-2016

2010-2016
Clinical treatment under compassionate use
GLP-certified laboratories at SR-TIGET

2016

CLINICAL TRIALS
supported by GXPs

2005
EMA ODD

2007
Protocol Assistance from EMA

2009
FDA ODD

2015
MAA filing at EMA

2016
MAA by EMA

REGULATORY ACTIVITIES