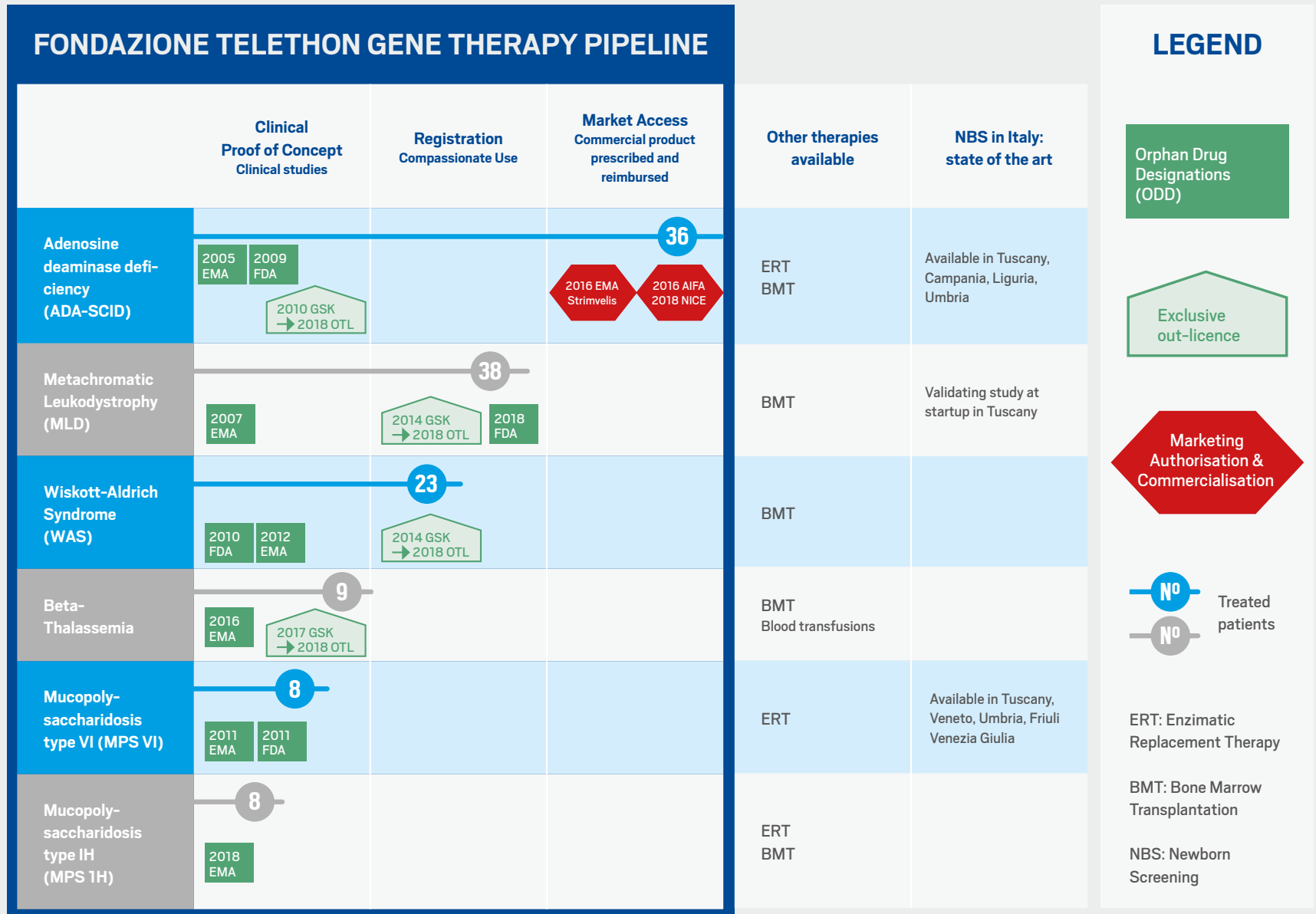


We made the therapies, now give us newborn screening!

ANNAMARIA ZACCHEDDU, ALESSANDRA CAMERINI, ERMANNO RIZZI (FONDAZIONE TELETHON, ITALY)

The development of gene therapy for genetic diseases is one of the most important results achieved by Fondazione Telethon. Such therapies could change the natural history of these diseases, especially if administered before the disease onset. How to perform an early diagnosis before the onset of symptoms? The newborn screening (NBS) is a health program that screens all newborn babies for many serious but treatable genetic disorders.

The NBS is also an essential tool to offer the available treatments to as many patients as possible and represents the only way to reach an early diagnosis to gain therapeutic benefits. Fondazione Telethon is at the forefront alongside patient associations to promote the culture of NBS and to sensitize institutions to extend it to an increasing number of diseases, given the new therapeutic options such as gene therapy. Strengthened by its leadership in the field of advanced therapies and media visibility, Fondazione Telethon has promoted strong communication and awareness-raising initiatives through all its channels.



Newborn Screening

Newborn screening is one of the most important preventive public health programmes.

A dedicated section on the website
www.telethon.it: 1,3 million users/year - 3,1 million impressions/year



In 2019, during the TV fund-raising marathon (> **10 million viewers/year**) 7 interviews have been focused on NBS
(photo: Prof Vincenzo Leuzzi and Laura, affected by phenylketonuria)



Caterina Balivo, a popular Italian presenter, realized a video to promote NBS during Rare Disease Day



We supported Leslie Martin, whose children Cajal and Ciaran are both affected by MLD, in his awareness campaign in Ireland. In 2019 he has been able to convince his government to enlarge the NBS panel, which counts only 7 diseases



Simon Harris TD
@SimonHarrisTD

New National Screening Advisory Committee set up. Brilliant group of people. Delighted to meet them today. Will review expansion of newborn screening programme as one of its 1st items of work

Traduci il Tweet



Articles on our quarterly magazine (> **850k issues/year**) and our social media platform (**Fb: 330k fans, Twitter 93k followers**)