Creating systematic and meaningful partnerships with the spinal muscular atrophy (SMA) community for therapy development

Fani Petridis,[1] Senam Beckley-Kartey,[2] Dylan Trundell,[3] Hannah Staunton,[3] Rakesh Kantaria,[2] Florian Lipsmeier,[4] Michael Lindemann,[4] Kristina Gelblin,[5] Ksenija Gorni,[1] Sangeeta Jethwa,[1] Tim Seabrook,[6] Omar Khwaja,[6,7*] Nicole Gusset,[8,9] Mencia de Lemus,[8] Vitaliy Matyushenko,[8] Inge Schwersenz,[8,10] Michael Ostland[2]

1. PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, Basel, Switzerland; 2. Pharma Development, Neuroscience, F. Hoffmann-La Roche Ltd, Basel, Switzerland; 3. Roche Products Ltd, Welwyn Garden City, UK; 4. Roche Pharma Research and Early Development, pRED Informatics, F. Hoffmann-La Roche Ltd, Basel, Switzerland; 5. F. Hoffmann-La Roche Pharma AG, Grenzach-Wyhlen, Germany; 6. Roche Pharmaceutical Research and Early Development, Roche Innovation Center Basel, Basel, Switzerland; 7. Voyager Therapeutics, Cambridge, MA, USA; 8. SMA Europe, Freiburg, Germany; 9. SMA Schweiz, Heimberg, Switzerland; 10. Deutsche Gesellschaft fuer Muskelkranke, Freiburg, Germany.

*Current affiliation.

Introduction

- Spinal muscular atrophy (SMA) is a rare genetic disorder characterized by progressive motor neuron loss which results in muscle wasting over time.
  - Increasing evidence suggests that SMA may also affect other types of cells and tissues throughout the body.
- Roche and Genentech are developing and testing risdiplam (RG7916), an investigational (unapproved) medicine developed in partnership with the SMA Foundation and PTC Therapeutics.
- Involving individuals with SMA and caregivers in our research leads to a better understanding of SMA as we investigate and develop potential therapies.

Roche wanted to make sure that:

- patient and caregiver voices are heard and integrated at all stages of the risdiplam clinical development program
- Roche communicates with the SMA community with one voice.

Building strong and lasting partnerships with the SMA community, required new ways of working.

Together with SMA Europe, Roche established a **standing advisory group** to promote **continued dialogue** and foster a **long-term partnership** with the SMA community.

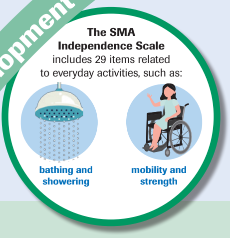
Roche
SMA Europe

Roche also appointed a **primary relationship manager** to **facilitate collaborations** across Roche functional teams, enabling the **integration of patient perspectives** across the life cycle of risdiplam.

Pre-approval

These initiatives created a streamlined and dynamic environment for mutually beneficial engagements between Roche and the SMA community. Examples of these engagements:

Clinical development


The SMA Independence Scale
includes 29 items related to everyday activities, such as:
bathing and showering
mobility and strength

Collaboration to develop patient-relevant trial outcomes

- Development of:
  - a **conceptual model of disease**, depicting the symptoms and impacts that are important to individuals with SMA and their families
  - the **SMA Independence Scale** to measure the amount of assistance that individuals with Type 2 and non-ambulatory Type 3 SMA require to perform daily activities
  - a **smartphone application** to capture changes in patient's day-to-day symptoms.

Communication of key updates to the risdiplam program

- Communicating transparently and continuously with SMA patient groups to make sure they have access to key program updates through:
  - community letters
  - FAQ documents
  - lay summaries of scientific publications
  - webinars
  - scientific advocacy briefings
  - family-friendly research posters.

Upon approval

Development of a patient-centric pre-approval access (PAA) program

- Partnering with the SMA Europe standing advisory group to ensure that the risdiplam PAA program was designed to provide equitable access to patients most in need when no other treatment options are available. Through this process we have been able to:
  - better understand the patient perspective on existing medical needs
  - redefine the inclusion and exclusion criteria and geographical breadth of the program to meet patient needs
  - broaden and/or validate ethical considerations to ensure equitable access and patient safety.

Roche is committed to involving and integrating patient and caregiver perspectives in all important program decisions. Partnering with the SMA community from the early stages of the risdiplam program in a systematic and consistent manner has been key to our risdiplam studies.

Acknowledgments

We would like to thank the patients and caregivers who participated in our research and SMA Europe for their ongoing partnership. The logos are the property of the companies listed. Editorial support was provided by Lindsey Weedon, of MediTech Media UK, funded by F. Hoffmann-La Roche Ltd. Genentech is a member of the Roche group.

Abbreviations

FAQ, frequently asked questions; PAA, pre-approval access; SMA, spinal muscular atrophy.

Presented at the 10th European Conference on Rare Diseases, Virtual Meeting, 15th–16th May 2020