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

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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.

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

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

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.

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

Clinical development

• 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.

• 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.

Communication of key updates to the risdiplam program

The SMA Independence Scale is included in the study protocol and is designed to measure all the functional changes that affect daily living. It includes both clinical and functional sections to allow for more meaningful and effective patient-reported outcome measures. It includes five levels of function: full (normal), significant (moderate), moderate (severe), severe (very severe), and nonsurviving. It is designed to allow for the collection of data on the impact of the disease on daily activities, including mobility, self-care, and social interaction. It is expected to provide valuable information on the effectiveness of risdiplam and its potential impact on the quality of life for individuals with SMA.

Acknowledgments

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.

Abbreviations:

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

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