Statement in support of the proposal on the inclusion of risdiplam in the WHO Model List of Essential Medicines for the treatment of spinal muscular atrophy

7 April 2023

Spinal muscular atrophy (SMA) is a rare neuromuscular disorder that is characterised by a progressive loss of motor neurons and a loss of motor and respiratory function, which usually leads to early death. The disease is diagnosed predominantly in small children. Approximately 60% of children born with SMA will experience an acute course of the disease – they will never acquire independent sitting or walking and will quickly deteriorate to the point of requiring 24/7 respiratory care and parenteral feeding. Life expectancy for children with this acute form is less than two years. A minority of children will experience milder progression; however, most will also lose much of their motor function with time.

SMA has been the leading genetic cause of death in infants and toddlers worldwide, and it remains so today everywhere where modern treatments are not available.

Modern treatments, introduced gradually from 2017 onwards primarily in high-income countries, work by stopping the loss of motor neurons. With an appropriate standard of care, treated patients experience meaningful improvements and many can expect normal life expectancy. The efficacy of these ground-breaking treatments is linked with the timing of diagnosis and therapy: the sooner the disease is diagnosed and a pharmacological treatment initiated, the higher the clinical benefit. If this occurs at the early, presymptomatic stage of SMA, most people receiving the therapy can expect a symptom-free life.

The typical incidence of spinal muscular atrophy is estimated at 1:10,000, even as it ranges from 1:11,200 in the United States, to 1:7,500 in continental Europe, to as much as 1:2,000 across the Middle East and North Africa. This translates into approximately **15,000 children being born with SMA in the world every year,** the majority of them affected by the fatal acute form.

As organisations founded and run by parents of children with SMA and people living with SMA, we would like to emphasise the stress and devastation that invariably accompany caring for a child or a family member affected by spinal muscular atrophy. This stress is always compounded by administrative and financial barriers to accessing a life-saving

therapy. The widespread misunderstanding and deprioritisation of rare diseases by many healthcare systems, combined with, frequently, high retail cost of treatments, remain an additional challenge globally.

Risdiplam is one the three modern treatments currently approved to treat SMA, and one with a remarkably patient-friendly route of administration. It is the only treatment that can be used by the patient at home. SMA patient organisations have played a significant role in its development, including funding preclinical research, consulting clinical study protocols, advising on the trial site selection, and leading on clinical trial enrolment. However, once approved, the therapy came with a prohibitive cost.

Since 2017, our organisations have campaigned for access to SMA therapies, including risdiplam. This dramatic struggle has brought mixed effects: while most (but not all) high-income countries have agreed to fund at least one SMA treatment through public healthcare, **no SMA treatments remain available or accessible in the vast majority of low- and mid-income countries as of 2023** despite more than 6 years passing from their introduction.

During that time, we have witnessed hundreds and thousands of children and adults dying a preventable death, and hundreds and thousands of families struck with unimaginable tragedies. With this in mind, **we would like to express our support to every initiative that has a potential to bring about easier, faster, and more equitable access to modern therapies and quality care for children and adults living with spinal muscular atrophy anywhere in the world.**

We remain at your disposal regarding any further discussion on spinal muscular atrophy, its effects and treatments.

(List of Signatories follows.)

List of Signatories

These signing Organisations represent and speak on behalf of SMA patient communities in their respective countries.

Become the Miracle Association (Taiwan)

www.smabma.org

Signed by: Li Yi Jie (Founder & Chairman)

"Children with SMA" Foundation (Ukraine)

www.csma.org.ua

Signed by: Vitaly Matyushenko (President)

Cure SMA Bangladesh

Signed by: Mohammad Shahadat Hossain (President) and Tanzina Afrin (Secretary).

Cure SMA India

www.curesmaindia.org

Cure SMA India has 1050+ patients registered from all over India. Its mission is to bring treatment and cure for SMA, making healthcare accessible, inclusive, and ensured for all.

Signed by: Alpana Sharma (Co-Founder & Director Patient Advocacy), Archana Panda (Co-Founder & Trustee)

Familias AME Argentina

www.fameargentina.org.ar

Signed by: Mariel Centurion (President)

Families of SMA Foundation (Hong Kong)

www.fsma.org.hk

Signed by: Mei Ling Fok, PhD (Founder & Chairman)

Family of SMA Russia

www.f-sma.ru

Signed by: Olga Germanenko (President)

Fundacja SMA (Poland)

www.fsma.pl

Fundacja SMA represents approximately 1,100 people with SMA and their families who live in Poland. It has successfully advocated for SMA treatments to become widely available in Poland.

Signed by: Kacper Rucinski (Co-founder), Katarzyna Pedrycz (Vice President)

Komunitas SMA Indonesia

www.smaindonesia.org

SMA patient support group of Indonesia founded in 2017

Signed by: Sylvia Sumargi (President)

Muscular Dystrophy Association (Singapore)

www.mdas.org.sg

Signed by: Sherena Loh, Director (Integrated Care & Special Projects)

Nätverket för spinal muskelatrofi (Sweden)

www.nsma.nu

Patient Voice Aotearoa (New Zealand)

www.facebook.com/patientvoiceaotearoa

Signed by: Fiona Tolich (Trustee)

Persatuan WeCareJourney (Malaysia)

www.wecarejourney.org

Signed by: Edmund Lim (President)

SMA Australia

www.smaaustralia.org.au

Signed by: Seona Donald (President)

Social Charitable Public Association "Genom" (Belarus)

www.facebook.com/myopathy

Signed by: Anna Gerina (Chair of the Management Board)

SMA Foundation Georgia

www.smaf.ge

Signed by: Guram Abuladze (Head) and Simoni Petridisi (Founder)

Spinal Muscular Atrophy Bhutan

www.facebook.com/SMABHUTAN

Although treatments are available elsewhere, they are not for us and others in Bhutan due to their unaffordability.

Signed by: Ugyen (Founder)

Spinal Muscular Atrophy Ireland Foundation

www.smaireland.com

Signed by: Jonathan W O'Grady (Director)

SMA Philippines Group

Signed by: Cathleen Po (Lead Advocate, Member of SMA APAC Alliance)

STOP SMA (Republic of Serbia)

www.stopsma.rs

Signed by: Marija Krstić (Founder)

TreatSMA (United Kingdom)

www.treatsma.uk

Signed by: Gennadiy Ilyashenko (Trustee)
