

## Statement

at the meeting of the 24th WHO Expert Committee on the Selection and Use of Essential Medicines,  
Geneva, 24<sup>th</sup> April 2023

by Kacper Rucinski

Ladies and gentlemen,

This week's meeting, here in the heart of green Geneva, is looked at with immense hope by thousands, tens of thousands of people around the world – people who have spinal muscular atrophy as well as parents of children with spinal muscular atrophy, like me.

People also not like me or like us, as we live in the privileged global North. That global North where the 20% of world's population have at their disposal 80% of resources. Where nearly all the ground-breaking, hyper-expensive therapies are readily available – but where they are also developed.

So, every two years, the global 80% pin their hopes on us here. And I'm here today with a task to speak on their behalf.

I'm a father of two girls, the younger of whom is 14 and has a so-called milder form of SMA. We are from Poland, my daughter was born and diagnosed in Georgia, spent time in Uzbekistan, we now live in the UK and have been taking part in a clinical trial of risdiplam in France. This has offered us a perspective, I guess. I have had a privilege of co-founding two SMA charities – SMA Foundation Poland in 2013 and TreatSMA UK in 2017 – and serving as a Board Member of SMA Europe. Today, I'm speaking on behalf of the 21 SMA organisations from all over the world who have signed a joint letter to the Committee about access to SMA therapies. My voice today is also joined by Julie Cini who had founded SMA Australia years ago and today is engaged in SMA patient advocacy across South-East Asia.

Firstly, what is SMA, or spinal muscular atrophy? SMA is a genetically determined neuromuscular disorder that manifests in infancy and early childhood in approximately 70% of cases – this is the so-called acute course. Otherwise healthy babies and young children suddenly stop achieving motor milestones and start getting weaker every week, every month. They are normal intellectually and emotionally; they just lose the ability to sit, to hold head, to lift hands. Their breathing grows weaker, they stop being able to swallow food... The end usually comes with a respiratory infection when their weak lungs are unable to clear secretions. Unless pharmacological treatment is quickly started, most children with the acute manifestation of SMA don't live past two years.

SMA has traditionally been called the biggest genetic killer of infants and small children.

People who have a milder manifestation, like my daughter, face a somewhat better perspective. Their disease progresses more slowly, and while they remain severely disabled and do get weaker, they don't usually die early. My daughter has been using a wheelchair since she was two. There are no words to describe the psychological burden when you realise that you, or your child, will only get weaker over time and will be forever dependent on other peoples' care.

To diagnose SMA is relatively easy – it's a simple genetic test that, in wholesale, costs below €2. Once diagnosis is there, the existing treatments, such as risdiplam, need to be started as soon as possible, as they stop the disease progress and often bring about significant improvement. Treated children learn to sit, many learn to walk. It's no longer a downward spiral. One can start planning their life, studies, work, relationships, because life now gains sense and perspective.

Let me stress that it was the SMA patient community that has funded the long development work of all the existing SMA therapies. Parents of severely ill children collected money to fund molecular scientists and clinicians. To-date, the SMA international patient community has collected and spent more than 160 million US dollars for the development of SMA therapies. It's owing to them that we now have three approved treatments.

However, even as more and more orphan drugs are receiving regulatory approval, we all know too well that rare genetic diseases are deprioritised by healthcare systems. Deprioritised in our countries, and even more deprioritised in lower-income countries. After all, common thinking is that anyone can get cancer, an infection, or get bitten by a rabid dog but having a genetic disorder must be rare. So, for some here may come as a surprise that there will be an estimated 1–2 SMA carriers in this room, and certainly many more if online participants are included.

One in 35 people in Europe is a SMA carrier. Twenty-one million people. If two carriers have a child, there's a 25% probability that the child will have SMA. The carrier rate indeed varies significantly – from approximately 1:40 in the US to 1:15 or more in the Middle East. Globally, around 15,000 children develop SMA every year: most of them, the acute form. At least 150,000 people in the world have SMA at any given moment. 80% of them in the global South where treatments are not available or affordable: one week of treatment costs a year's wages.

Quick calculations show that before this meeting ends on Friday, 150 children in low- and middle-income countries will die of SMA. If the Committee decides to delay the decision to 2025, it will cost the life of additional 20,000 children.

I ask you not to think of rare diseases like SMA as a marginal issue. Please see them as an urgent, deprioritised global health topic where only this committee is in a position to bring about a meaningful change.

Thank you. On behalf of the 80%.

Kacper Rucinski

*Co-founder and Strategy Board Member, SMA Foundation Poland*

*Co-founder, TreatSMA UK*

*National Delegate, SMA Europe*

*Member, Steering Committee, European Alliance for Newborn Screening in Spinal Muscular Atrophy*

*Member, Steering Group, UK SMA Newborn Screening Alliance*

*Owner, Rucinski Consulting*