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Risdiplam (Evrysdi) in the treatment of spinal muscular atrophy – a new therapeutic perspective in Poland

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Abstract

Spinal muscular atrophy (SMA) is a rare autosomal recessive neuromuscular disorder caused primarily by mutations or deletions in the *SMN1* gene, resulting in survival motor neuron (SMN) protein deficiency and progressive motor neuron degeneration. Over the last decade, the introduction of disease- modifying therapies has substantially altered the natural course of the disease. This review article presents the pathophysiology of SMA, discusses available therapeutic options, and provides a comprehensive overview of risdiplam, an orally administered *SMN2* splicing modifier. The paper summarizes its mechanism of action, pharmacokinetic and pharmacodynamic properties, and key clinical trial data. Long-term safety outcomes and the role of risdiplam in the Polish healthcare system following its reimbursement in October 2024 are also discussed. Particular attention is given to implications for pharmaceutical practice, including patient education and adherence monitoring. This article also analyzes the major social and economic costs of living for patients and their families during long-term treatment. *(Farm Współ 2026; 19: 88-96) doi: 10.53139/FW.20261914*

Keywords: spinal muscular atrophy, risdiplam, SMN2 splicing modifier, disease-modifying therapy, newborn screening, Poland

Introduction

Spinal muscular atrophy (SMA) is a severe, degenerative α-motor neuron disease with autosomal recessive inheritance that impairs neuromuscular development, leading to weakness of skeletal and respiratory muscles and joint contractures [1]. It is a genetic disorder that, in 95% of cases, results from a homozygous deletion or a loss-of-function mutation in the *SMN1* gene on chromosome 5q11.2-13.3 [2]. A deficiency of the SMN (Survival Motor Neuron) protein induces hypotonia, gradual muscular atrophy, breathing difficulties, and physical impairment, often leading to disability or death [3].

SMA is considered a rare disease, the second most common autosomal recessive disorder [4]. Moreover, it is believed to be the leading factor responsible for infant mortality. SMA occurs in approximately one in 10,000 to 20,000 live births, and its carrier frequency in the general population ranges from 1/40 to 1/70 [1]. Epidemiological data are scarce, although estimates range from 800 to 1000 SMA patients in Poland. SMA is diagnosed in 50-60 children in Poland every year, of whom 35-55 will develop acute SMA, and symptoms will appear in infancy [3]. However, the introduction of disease-modifying therapies has significantly transformed the prognosis of SMA, shifting it from a primarily fatal pediatric disorder to a treatable chronic condition.

Clinical classification of SMA

SMA results in a wide range of clinical severity, depending on the age at which the first symptoms appear and the ability to achieve milestones in motor development, including sitting, standing, and walking independently. Nevertheless, some symptoms are constant regardless of how the illness progresses, such as the following: 1) symmetrical, progressive muscle weakness - the weakest are the muscles of the pelvic girdle, followed by the muscles of the shoulder girdle, 2)

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preserving the diaphragm and the external muscles of the eyeball, 3) low muscle tone, 4) weakening or loss of deep reflexes - the first reflex to disappear is the patellar reflex, 5) finger tremors. However, there are no pyramidal symptoms, sensory or sphincter abnormalities, or altered psychological development [5].

SMA is divided into various phenotypes (SMA types 0 to 4) based on age of onset and the severity of the clinical symptoms, which are inversely correlated with the amount of SMN protein present at the motor neuron level. The number of copies of *SMN2*, a paralogue of *SMN1* that produces low levels of functional SMN protein and can partially compensate for the absence of *SMN1*, determines the disease's phenotype--genotype correlation [6].

The classification of clinical types is presented in the table I.

The most severe form of SMA is type 0. It usually manifests during pregnancy and causes death within a week after delivery. These individuals have reduced fetal movement in utero, joint contractures, poor feeding, hypotonia, and a feeble cry. At the time of birth, patients with type 0 SMA usually require respiratory support [7]. Type 1 SMA, also called Werdnig-Hoffman disease, manifests as limb weakness, weak crying, respiratory distress, poor feeding, tongue fasciculation, and a "frog-log" posture between birth and 6 months of age. Sitting with assistance is the maximum level of motor function attained, and patients frequently pass away from respiratory failure at less than the age of two [7]. Most children with SMA type 2 can sit on their own but can never walk, and symptoms often appear between 6 and 18 months old. They typically have difficulty with swallowing, which can lead to weight loss. Moreover, patients develop kyphosis, fine hand tremors, and tongue fasciculation. The majority of them will die before turning 25 [8]. Kugelberg-Welander disease, another name for SMA type 3, is a milder variant that typically manifests later in childhood, generally beyond 18 months. Patients may initially be able to walk unassisted, but they usually develop severe scoliosis. They frequently lead normal lives but lose the ability to walk at a young age [9]. The last type of SMA is type 4, often known as adult-onset SMA. Throughout the course of the illness, these patients can walk on their own and have very moderate muscular issues. A patient with type 4 SMA will often live a normal adult life [7].

Therapeutic options of treatment

For many years, till 2016 when the FDA approved the first medication, the treatment was mainly symptomatic. Classically, palliative care has been used to manage SMA, addressing difficulties as they arise [10]. Since the primary cause of death in SMA is respiratory failure, the supportive treatment includes mechanical ventilation, involving invasive ventilation for the most severe cases and noninvasive ventilation and tracheostomy for the milder types of SMA [8]. Rehabilitation, occupational therapy, and physiotherapy all help preserve muscle strength, prevent contractures, and increase range of motion. Walking aids, wheelchairs, braces, and orthoses help to enhance functional independence [11]. Some patients need additional therapy for speech and swallowing difficulties. Furthermore, these individuals need a healthy diet and adequate

Table I. The clinical classification of SMA types based on the average number of *SMN2* copies, the percentage of patients, and the natural course of the disease [3]

SMA type	Average number of *SMN2* copies	Percentage of patients	Natural course of the disease
Type 0	1	Rarely, less than 1%	Death within a few weeks after birth
Type 1	1-2	45-60%	Lifespan of <2 years
Type 2	3	20%	Life expectancy is shorter than in healthy people (estimated to be 20-40 years)
Type 3	3-4	30%	Lifespan the same as in the case of healthy people
Type 4	4 or more	Less than 5%	Lifespan the same as healthy people

caloric intake to maintain muscle mass and weight while avoiding prolonged fasting. A feeding tube may need to be inserted for people who are unable to chew or swallow [12]. As mentioned before, the therapy was focused on the treatment of complications, but it did not modify the disease's progression. However, major medical and scientific advances over the past 10 years have significantly improved the quality of life and life expectancy [13].

The natural course of SMA has been significantly altered by the introduction of disease-modifying treatments that increase SMN protein levels, including gene therapies and *SMN2* splice modulators [14]. The first drug specifically developed to treat SMA patients with all disease types is intrathecal nusinersen (Spinraza), which increases the amount of full-length SMN protein. It was approved by the FDA in December 2016. In May 2019, the FDA approved onasemnogene abeparvovec (marketed under the brand Zolgensma) for the treatment of children younger than 2 years old with a diagnosis verified by genetic testing. Zolgensma is a one-time intravenous injection that induces the long-term production of full-length SMN protein [15].

Lastly, risdiplam (Evrysdi) was approved by the FDA in August 2020; however, it was not included in the Polish drug program until October 2024 [16]. Evrysdi is the first and only oral, non-invasive treatment for SMA that is an *SMN2* splicing modifier. The major benefit of this medication is its previously mentioned oral administration, which can affect systemic tissues implicated in the multisystem pathophysiology of this disease [7]. Patients and their families can receive therapy at home, which increases their comfort level. Introducing risdiplam marks a significant advancement in the personalization of the treatment for each individual who is facing spinal muscle atrophy.

Risdiplam's mechanism of action

As mentioned above, in humans, the SMN protein is encoded by the *SMN1* gene, which produces most of the functional motor neuron SMN protein, and the *SMN2* gene. Only 10-25% of the *SMN2* mRNA molecules are the source of the full-length, effective SMN protein, as the pre-mRNA of the *SMN2* gene undergoes distinct post-transcriptional processing, resulting in most of the mRNA being exon 7-deleted, and the produced SMN protein being shorter, non-functional, and quickly degraded [17].

Hence, risdiplam is a small-molecule splice modulator of the *SMN2* gene. It directly interacts with *SMN2* pre-mRNA and targets two different locations: the 5' splicing site of exon 7 and an exon enhancer sequence.

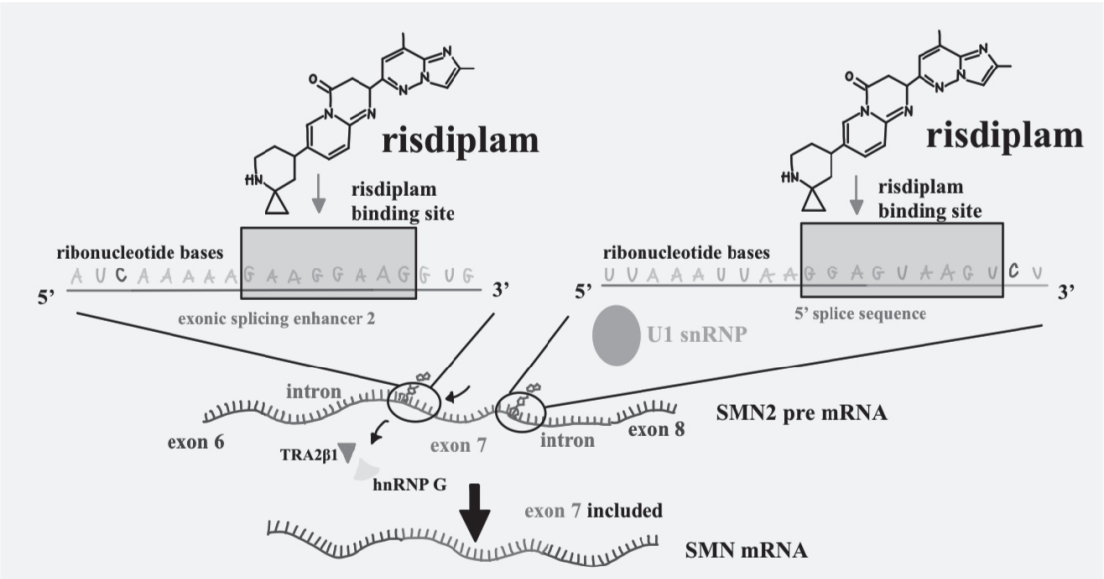

Figure 1. Risdiplam structure and mechanism of action [19]

The ribonucleoprotein complex- U1 snRNP is stabilized as a result of this interaction, which makes it easier to include exon 7 and increases the production of full-length SMN protein [17]. Infants treated with risdiplam for a full year had considerably longer event--free survival than expected for the disease's natural course. Risdiplam increased SMN protein levels from baseline by more than 2-fold within 4 weeks of starting treatment, and these effects persisted for at least 1 year [18]. Figure 1 below presents the risdiplam structure and mechanism of action [19].

Pharmacokinetics and pharmacodynamics of risdiplam

Risdiplam's pharmacokinetics were roughly linear between 0.6 and 18 mg in healthy adults and between 0.02 and 0.25 mg/kg once a day in patients with SMA following oral Evrysdi treatment. When risdiplam was administered orally once daily to healthy subjects, peak plasma concentrations (C_{max}) and the area under the plasma concentration-time curve ($AUC_{0\text{-}24h}$) increased approximately 3-fold. After daily dosing, risdiplam exposure reaches a stable state within 7 to 14 days [20]. Risdiplam is rapidly absorbed when administered on an empty stomach; T_{max} in plasma after oral administration is 1-5 hours. Food (high-calorie/ high-fat meal) does not significantly affect the exposure of the medication [21]. Risdiplam is evenly distributed to all parts of the body, including the CNS, by penetrating the blood-brain barrier. It is mainly bound to serum albumin, not bound to alpha-1 acid glycoprotein, and the free fraction is 11%. It is metabolized mainly by FMO1 and FMO3, as well as by CYP1A1, CYP2J2, CYP3A7, and CYP3A4 [21]. The effective $t_{1/2}$ of the medication in healthy adults is approximately 50 hours. Around 53% of the dose is excreted in the feces and 28% in the urine. The parent compound is the major component in plasma, accounting for 83% of the total drug-related substances in the bloodstream [20].

Risdiplam's drug information

It is advised to take Evrysdi orally at approximately the same time every day with or without food. It comes in two forms- liquids and tablets. Age and body weight dictate the recommended dosage. Evrysdi tablets offer flexibility to the community, thanks to refrigeration-free storage and simplified dosing, as each tablet contains the patient's dose. However, patients must be ≥2 years old and weigh ≥20 kg [20]. After the liquid medication is drawn into the oral syringe, it should be taken immediately. The administration should be followed by drinking water [7]. If a dose is missed, it should be taken as soon as possible, ideally within 6 hours of the missed dose. The next day, the regular dosing plan should be resumed. Otherwise, the missed dose should be avoided. [20]. If the medication is not completely swallowed or vomiting occurs, it is not advised to take another dose to make up for the missed dose [7]. According to in vitro research, Evrysdi may raise the plasma concentrations of medications such as metformin that are removed by MATE1 or MATE2-K. The coadministration of Evrysdi with MATE substrates should be avoided. Furthermore, mild and moderate hepatic impairment does not significantly affect the pharmacokinetics of risdiplam [21]. Data about risdiplam use in pregnant women is insufficient. Animal research has shown reproductive toxicity and potential teratogenic effects on the embryo and fetus [22].

Long-term safety data

Long-term extension studies show a permanent increase in SMN protein levels in the blood, along with prolonged stabilization or improvement in motor performance. The majority of risdiplam's side effects are low to moderate in severity, and the drug's safety profile has not changed over time [23]. In 10% of patients with later-onset SMA, the most frequent adverse effects of risdiplam are fever, diarrhea, and rash. The adverse effects of infantile-onset SMA were similar to those of later-onset SMA, with the addition of 10% of patients experienced vomiting, constipation, pneumonia, and upper respiratory tract infections. Urinary tract infections, arthralgias, and mouth and aphthous ulcers are less frequent adverse effects of risdiplam in later-onset SMA [7].

Preclinical research conducted in mice and non--human primates addressed nusinersen's bioavailability issues by confirming risdiplam's effectiveness in restoring appropriate SMN protein levels. In fact, several preclinical investigations have shown that the SMN protein performs a wide range of functions in many tissues and at various levels, which explains the pathology's non-motor symptoms [24]. Nevertheless, comprehensive real-world evidence data, particularly from the Polish population after reimbursement in 2024, remains limited and will be crucial for evaluating long-term safety [6].

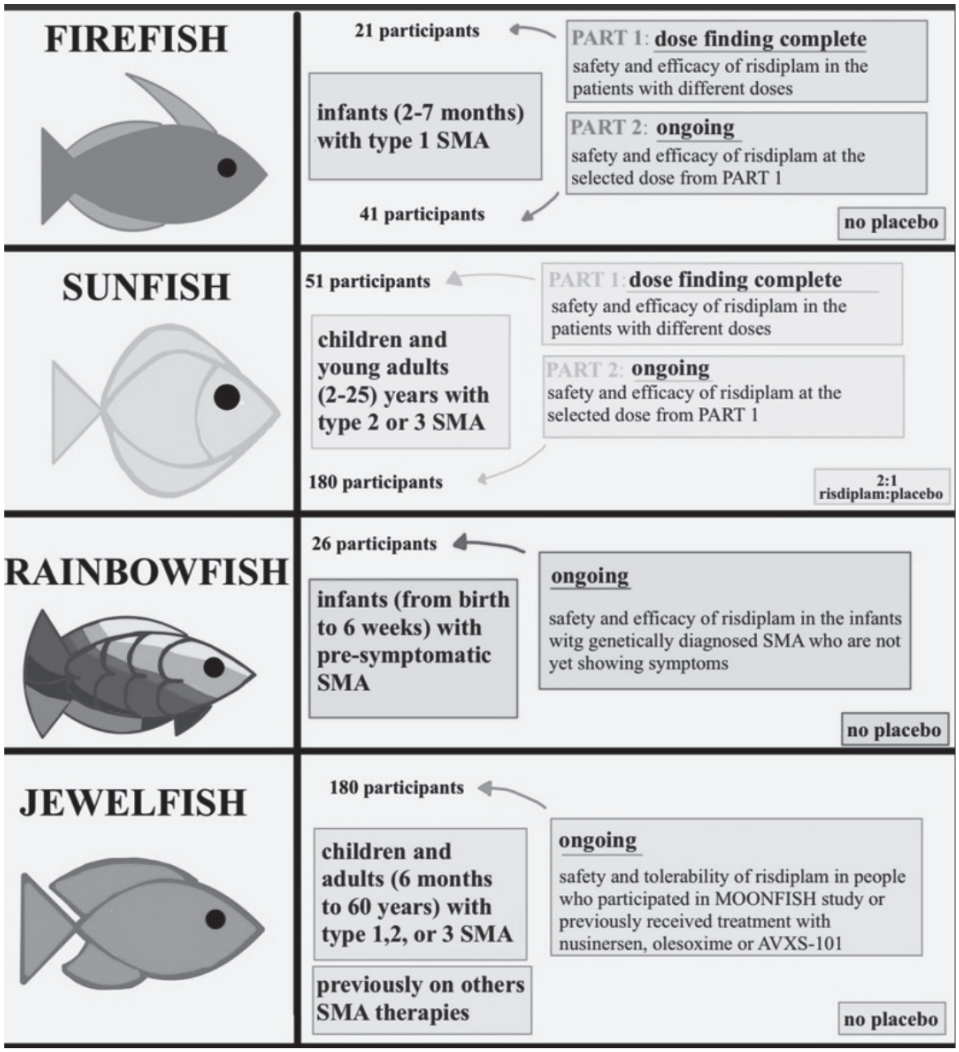

Figure 2. The risdiplam clinical trial program [25]

Clinical trials

More than 490 individuals aged from newborn to 60 years with pre-symptomatic Type 1, 2, or 3 SMA were enrolled in the Evrysdi clinical trial program. A comprehensive research program includes trials shown in Figure 2 [18,25].

FIREFISH Trials

The purpose of these two-part open-label FIREFISH trials was to establish Evrysdi's safety, tolerability, pharmacokinetics, and pharmacodynamics, as well as the drug's effectiveness and how the body reacts to it. The drug's efficacy was established from outcomes measured at 12 and 24 months of treatment, as presented in table II. The BSID-III scale was used to assess the ability to sit, which, in the natural history of infantile-onset SMA, would not be expected to occur. Moreover, FIREFISH trials also assessed additional motor milestones, such as head control, rolling, voluntary grasp, SMN protein levels in blood, and the percentage of infants who could be fed orally [18].

SUNFISH Trials

The tolerability, safety, effectiveness, pharmacokinetics (PK), and pharmacodynamics (PD) of Evrysdi were evaluated in this two-part clinical trial. In Part 2,

Table II. Clinical outcomes in the FIREFISH Trials after 12 and 24 months of treatment

Outcome	12 months	24 months
Sitting ≥5 s	33%	60%
Sitting ≥30 s	–	40%
Standing	–	28%
Alive without permanent ventilation	87%	84%

Table III. Clinical efficacy outcomes of risdiplam in the SUNFISH Trials at month 12

Outcome (Month 12)	Evrysdi	Placebo
Mean change in MFM-32 score from baseline	+1.36	-0.19
Patients with ≥3-point improvement in MFM-32	38%	24%
Upper limb motor function (RULM)	Improvement compared to baseline	

Note: MFM-32– Motor Function Measure, RULM – Revised Upper Limb Module.

Table IV. Clinical outcomes in the RAINBOWFISH Trials in presymptomatic infants treated with risdiplam

Outcome	12 months	24 months
Sitting without support ≥5 s	96%	100%
Sitting without support ≥30 s	81%	91%
Standing	84%	96%
Walking independently	48%	87%
Alive without permanent ventilation	100%	–
Exclusive oral feeding and swallowing	100%	–

Note: Motor milestones were assessed using the Bayley Scales and Toddler Development (BSID-III) and the Hammersmith Infant Neurological Examination Module 2 (HNE-2).

180 adults and children were randomized 2:1 to receive Evrysdi or a placebo. The main efficacy results at month 12 are summarized in table III [26].

RAINBOWFISH Trials

The efficacy of risdiplam was also evaluated in the open-label RAINBOWFISH trial in newborns with presymptomatic SMA. Considering the infantile--onset natural history, untreated infants with type 1 SMA never learn to sit without assistance. The study included 26 infants. Key motor milestones and survival outcomes are summarized in table IV [27]

Apart from FIREFISH, SUNFISH, and RAINBOWFISH, Evrysdi is also participating in a trial called JEWELFISH, which assesses SMA patients aged 6 months to 60 years who have received various approved or experimental SMA treatments for at least 90 days before starting Evrysdi [18]. Risdiplam is thus a drug with proven long-term effectiveness that can be used for any type of SMA in patients of all ages.

Importance in pharmaceutical practice and the Polish Health System

The significance of Evrysdi in Polish pharmaceutical practice primarily concerns its availability and reimbursement. As of October 1, 2024, risdiplam was included in the drug program, which enabled all patients to receive treatment under the public system [16]. By coordinating access, monitoring therapy, and providing guidance on appropriate use, pharmacists are essential to managing the clinical and financial challenges of SMA therapies [28]. They play a key role in educating patients and their families about dose guidelines and potential adverse effects, in monitoring compliance with therapeutic recommendations, and in reporting any adverse reactions. Furthermore, pharmacists assist with pre-treatment evaluations, including laboratory and antibody testing relevant to gene therapy eligibility [29]. Additionally, they collaborate with specialized centers that treat SMA, helping to optimize therapy and participating in the process of registering clinical data [30].

A significant milestone in Polish SMA care was the implementation of a nationwide newborn screening program in 2021, enabling genetic diagnosis before symptom onset [31]. The screening program's data demonstrate rising detection rates and early therapeutic intervention, both of which are essential for optimal results [32]. Polish centers have actively participated in global clinical trials, including FIREFISH and SUNFISH, contributing substantially to patient recruitment and accelerating drug development [16]. Despite progress in current medical practice for SMA, there are still significant challenges to overcome, especially regarding the high cost of medication and the requirement to maintain long-term access to the drug under the program [33].

Socioeconomic Cost of Living with SMA

Notwithstanding advancements in therapy, living with SMA comes with substantial social and financial expenses. Both direct medical expenses and significantly higher non-medical or societal expenditures are part of the financial burden of SMA [34]. Direct costs include expenses related to the patient's treatment and medical care. The purchase of medical equipment (ventilators, wheelchairs, adaptive strollers, or orthoses, etc.), regular hospitalizations and specialists' consultations, or frequent physical therapy are the most crucial of these [35]. The introduction of the oral form of the medicine relieves strain on healthcare institutions, allows therapy to be administered at home, and reduces the need for hospitalization [27]. Nonetheless, limitations in patients' and caregivers' professional activities are the main driver of indirect costs. In the case of children who suffer from severe forms of SMA, one of the parents frequently needs to quit their job or drastically cut back on their job duties to provide the child with continuous care. As a result, there is a greater chance of financial problems and a partial loss of household income [36]. According to available data, in Poland, only about 30% of caregivers work professionally [37]. The decline in a patient's future economic potential is another example of an indirect cost, particularly when the illness begins in early childhood and affects education and the ability to pursue employment later in life. For the family of patients, the illness is linked to a significant emotional and social burden in addition to financial ones. It requires considerable effort and time to care for someone with a chronic disease, which can result in ongoing stress and caregiver exhaustion. Families frequently have to rearrange their daily routines to accommodate a sick child or adult [38]. Patients' limited mobility and the need for specific equipment may also make it more difficult for them to participate in social and educational activities, leading to social isolation [39]. In Poland, there is an organization called Fundacja SMA that supports individuals with SMA and their families. It plays an important role in improving the quality of life of those affected and increasing public awareness of SMA. Moreover, this group facilitates access to rehabilitation, conducts conferences and family meetings that enable the exchange of experiences, and assists families in learning about available treatments. Additionally, the Fundacja SMA actively collaborates with public institutions and medical experts worldwide to improve access to modern therapies and diagnostics [40]. The Fundacja SMA's initiatives show that non-governmental groups can have a real and measurable impact on the lives of people affected by rare diseases; therefore, it is crucial to support them to develop a patient-centered and comprehensive system of care for people with these severe conditions.

Conclusions

Spinal muscular atrophy has changed from a rapidly progressive and frequently fatal pediatric disorder to a chronic condition that can be treated due to the development of disease-modifying treatments. Targeted therapies that raise SMN protein levels and dramatically change the disease's natural course have been made possible by advances in molecular medicine, especially when treatment is started early.

Risdiplam represents an essential component of the current therapeutic environment. Its systemic distribution and oral route of administration set it apart from other available treatments and enable the restoration of SMN protein in both central and peripheral tissues. Clinical trials across a wide variety of SMA phenotypes and age groups have shown a generally positive long-term safety profile, sustained increases in SMN protein levels, and significant improvements or stabilization of motor function.

In Poland, the full reimbursement of risdiplam since October 2024 and the countrywide newborn screening program have greatly expanded therapeutic access and opened new possibilities for early, individualized treatment strategies. Despite the relatively small number of patients in Poland, estimated at approxima-

tely 1,000, SMA has become a significant illustration of how patient advocacy, scientific advancement, and activism can influence the development and accessibility of innovative therapies. Raising public awareness of rare diseases can be extremely important for promoting investment in novel medicines, accelerating research, and expanding access to currently available treatments. The transition to home-based oral therapy also strengthens pharmacists' role in patient education and pharmacovigilance and highlights the importance of interdisciplinary teamwork. Despite advances in treatment, the financial, social, and psychological challenges associated with the disease remain considerable. Even with promising results from clinical trials, continued collection of real-world evidence in the Polish population remains essential. Long-term observational data and national registries will be crucial for assessing the durability of response, safety in routine clinical practice, and the overall impact of risdiplam on patient quality of life. Further integration of clinical, pharmacological, and registry data will help optimize treatment algorithms and support evidence--based decision-making in the evolving management of spinal muscular atrophy.

Conflict of interest
None

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