What is SMA?

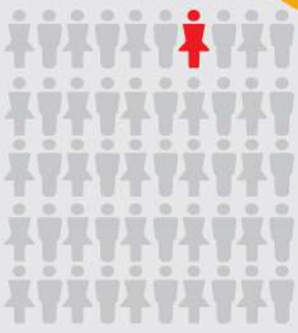
Quick facts about spinal muscular atrophy (SMA)

In Malaysia, it is estimated that **one baby** with SMA is born every week.

SMA is an inherited genetic disease.

It occurs when a child inherits **two mutated copies of the survival motor neuron 1 (SMN1) gene** – one from each parent (autosomal recessive inheritance).

If a child only inherits one mutated copy of the SMN1 gene, he or she becomes a **carrier** of the recessive gene.



On average, **one in 50 people** carry the mutated SMN1 gene.

SMA is a neuromuscular disorder, affecting the motor nerves in the body.

It causes the **muscles** in the body to become **progressively weaker**.

It affects **essential physical activities** like breathing and chewing/swallowing, as well as all movements, including head control and crawling/walking.

SMA does not affect the child's intelligence or senses.

SMA can be categorized into four clinical types:

SMA Type 1

> Most common and severe type.
> Symptoms appear at birth or within first few months of life.
> Patients have difficulty breathing and eating.
> Usually do not live beyond two years of age.

SMA Type 2

> Symptoms appear between six to 24 months of age.
> Patients can sit by themselves, but are unable to stand or walk on their own.
> Life expectancy ranges from childhood to adulthood, depending on the severity of their condition.

SMA Type 3

> Symptoms appear during childhood.
> Patients are initially able to walk, but will eventually need a wheelchair.
> Life expectancy is close to normal.

There is **no cure** for SMA, but there are **therapies** to **slow the progression** of the disease and **relieve some of the symptoms**.

SMA Type 4

> Symptoms appear during adulthood.
> Patients are initially able to walk, but will experience progressive muscle weakness.
> Life expectancy is normal.

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How SMA affects me physically

Here are five physical limitations that affect persons with spinal muscular atrophy (SMA) the most on a daily basis.

"I am not worried about death. I am worried about being an invalid."

– A woman in her 30s with SMA type 2

"In this study, the foremost issue was the lack of independence and mobility. In particular, this was a critical milestone for patients with SMA type 3 as they transitioned from being independently mobile to becoming wheelchair-bound. On the other hand, wheelchairs are a part of life for those with SMA type 2, who are usually adept at utilising powered or electric wheelchairs. However, all face difficulty travelling independently or navigating their environment due to sub-optimal infrastructure and public amenities that are inaccessible to wheelchairs."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

1. Being unable to move about independently

2. Being unable to transfer one's self from one piece of equipment to another (e.g. from wheelchair to bed)

3. Being unable to conduct normal physical activities

4. Being unable to go to the toilet by themselves

5. Unable to be truly independent

"My most worrisome situation is when I suddenly have phlegm in my lungs [and] I cannot cough it out and the helper is not able to push or rub my body correctly to expel it. Another major concern I can think of maybe is deterioration in respiratory function. I may not be able to take it as I am scared and also worry too much over this declining respiratory function. I am concerned that when the time comes, I may need to rely upon and be dependent on a respirator when my respiratory system deteriorates."

– A woman in her 40s with SMA type 2

The main symptoms of SMA that affect a patient's quality of life are:

Muscle weakness

Joint contractures

Lung infection

Fatigue

Inability to cough effectively

"Because with SMA, you get weaker with time – progressive weakness – I worry about losing the ability to do things I like and the things that matter, for example, just being able to hold the fork and spoon. ... It is something basic, but I am losing my ability to do it, so I worry about all this."

– A man in his 20s with SMA type 2

"My son was not achieving his milestones, like crawling or sitting up by himself, by the time he reached six months, unlike his older brother. He had a very soft cry and soft limbs. He couldn't lift up his head well. The doctor called it hypotonia. We did physiotherapy for six months but it didn't help."

– A mother of a boy with SMA type 1

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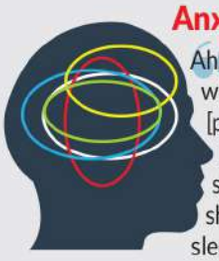
How SMA affects us mentally

The mental health of persons with spinal muscular atrophy (SMA) and their caregivers can be significantly impacted by the presence of the condition.

DIRECT IMPACT

Anxiety



"Ah, at night, I never slept soundly. Every once in a while, I would get up to pray. I would recite [prayers] with my husband. I sleep, I recite, we look at her – we look at her every night. I can't sleep because I am scared [that] when I wake up, she won't be here because ... Every night I can't sleep (voice chokes with emotion)."

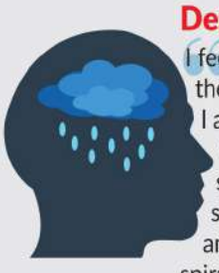
– A mother of two deceased children with SMA type 1

Stress

"Women, when we return home after work, we will be stressed – stressed from work and bring it home. [Then] at home, I will have to handle [the chores]. Sometimes, I feel burnt out."

– A mother of two children with SMA

Depression



"I feel [frustration and hopelessness] most of the time, I don't know why. Maybe because I am alone, so I get overwhelmed with everything. Maybe it is too much to bear sometimes ... I also have my limits, sometimes I keep pushing myself and I am like: AAHHH! And just, you know, spiraling into despair."

– A woman in her 20s with SMA type 3

Grief (for caregivers)

"Some people commented, 'Don't worry, you're still young, you can get pregnant again.' It's very hurtful, you know?! No one can replace anyone. Even if he is no longer here, even if I have 10 babies, 10 more kids, we cannot replace this baby. Even now, with our second child, we talk, we tell him, you had a big brother before."

– A mother of a deceased child with SMA type 1

"There is need for a support system at each stage of SMA as many parents have painfully shared the mental torment, anger, stress, depression and anxiety they go through at each stage of the disease. This includes dealing with the shock and grief that comes with the diagnosis and prognosis, the mental burdens of caring for their child and the grief when the child passes away."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

INDIRECT IMPACT

On the parents' relationship

"The differences between male and female ways of handling and expressing emotions such as non-acceptance, anger, guilt and fear, can also potentially lead to a rift in the marriage. ...

The hereditary factor of genetic diseases like SMA can pose unique issues for patients and families. This includes parental guilt regarding transmission, the potential for prenatal diagnosis, ethical and moral dilemmas with regards to the ability to terminate an affected pregnancy, and foreknowledge of the disease and its prognosis."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

On siblings

"The care of a child with SMA means that other children (their siblings) are given less attention, and therefore, often felt neglected. Mrs SS [a mother of a boy with SMA type 1] shared the experience of how her older child started hurting himself when he was four-years-old, in order to get her and her husband's attention."

Living With Spinal Muscular Atrophy in Malaysia: A Study

On social and family relationships

"For family occasions, kenduri, weddings, I would reject invitations from all my relatives; I never went for any."

– A mother of a deceased child with SMA type 2

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How much SMA costs

Below are the various expenses related to the management of spinal muscular atrophy (SMA), which can greatly strain a family's finances.

HEALTHCARE

As SMA involves progressive weakness of the body's muscles, many medical specialists and allied health professionals can be involved in the care of a person with SMA (PWSMA). These include:

- Paediatrician
- Geneticist
- Neurologist
- Respiratory physician
- Orthopaedic surgeon
- Gastroenterologist
- Physiotherapist
- Occupational therapist
- Nutritionist/Dietitian
- Speech therapist

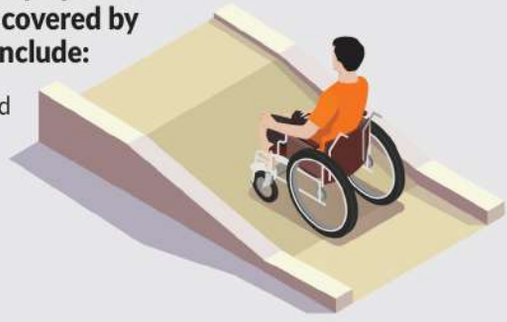
There are currently three drugs that can treat SMA: nusinersen (Spinraza), onasemnogene abeparvovec-xioi (Zolgensma) and risdiplam (Evrysdi).*

While healthcare services and medications can be obtained at a minimal cost at government hospitals, the long waiting periods and short time availability of healthcare professionals may drive PWSMA and their caregivers to seek private services.

EQUIPMENT

PWSMA often require crucial equipment to help manage their symptoms that are neither provided for by the government hospitals nor covered by medical insurance, and must be paid for out of pocket. These include:

- Cough assist devices
- Non-invasive ventilation, e.g. CPAP and BiPAP
- Suction devices
- Orthotics
- Mobility devices, e.g. powered wheelchair
- Nutritional supplements
- Home renovation, e.g. to fit wheelchairs



OTHER EXPENSES

- Obtaining and paying for a **domestic helper** (usually foreign workers), especially for independent adult PWSMAs.
- **Travel and accommodation** while obtaining healthcare services only available far from the PWSMA's home.

*(Note: This might also entail **loss of income** for PWSMAs or caregivers paid by job or time.)*

Couples who are both carriers of the mutated SMN1 gene that can result in SMA may also wish to undergo:

- Prenatal diagnosis (to determine if their fetus has SMA)
- Pre-implantation genetic diagnosis (to determine if their embryo – formed through assisted reproductive technology – has SMA)

Each of these procedures has to be paid out of pocket by the couple.

POLICY RECOMMENDATIONS

- **Subsidise the drugs** that can treat SMA if and when they are approved in Malaysia.
- Implement **a systemic approach to preventing and diagnosing SMA** at a population level, i.e. prenatal diagnosis, newborn screening and pre-implantation genetic diagnosis.
- **Remove taxes** on imported medical equipment essential for the management of SMA.
- **Prohibit genetic discrimination** by insurance companies.
- Create a **respite care programme** for caregivers of PWSMA.

**None of them are currently available on the market in Malaysia.*

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Education, employment and social inclusion

Persons with spinal muscular atrophy (PWSMA) have and are encountering barriers in these three areas that prevent them from achieving their full potential.

> The ability to lead a normal life is particularly imperative for parents with adult SMA children and the PWSMA themselves.
>
> – *Living With Spinal Muscular Atrophy in Malaysia: A Study*

BARRIERS TO EDUCATION

➢ The misconception that PWSMA are intellectually challenged.

> One common experience had by both PWSMA and caregivers is the assumption on the part of education officials that a child with a physical disability should be placed in the special education class (i.e. due to learning disability), even though the risk of learning disabilities for PWSMA is the same as the general population.
>
> – *Living With Spinal Muscular Atrophy in Malaysia: A Study*

➢ Discrimination against persons with disabilities

> She said they couldn't have [my daughter] unless I have someone or a full-time PA (personal assistant) look after her. The teacher reasoned that if she assists [my daughter], the class would stop midway, disrupting it. The teacher even mentioned that the PIBG had complained that it would interrupt the class if [my daughter] continues to be there. I was shocked! So shocked! I felt like it was not fair for my kid, but if I let [her] stay in that school, I was afraid they might do something to my daughter, so I took her out from that school immediately.
>
> – *A mother of a woman with SMA type 3*

➢ Lack of understanding and compassion

> I think that primary school was the hardest time of my life because I faced many challenges. For instance, my classroom was on the first floor and the teachers weren't very ... I would say, supportive. My parents talked to them, but they still didn't put my class downstairs, so I had to climb up the stairs. Sometimes I was so tired and my dad had to carry me up.
>
> – *A man in his 20s with SMA type 2*

➢ Bullying

> There was one incident (long silence) of bullying because ... because of my condition, I couldn't lift my head up. If I fall down now, I can't lift my head up. Yeah, so ... my classmates when I was in primary school in normal class ... one of them pushed my head down ... and ... and pressed. Well, of course, as my head was down, I couldn't lift it up (silence). Yeah ...
>
> – *A man in his 20s with SMA type 2*

BARRIERS TO EMPLOYMENT

➢ The misconception that PWSMA are intellectually challenged

> One common experience had by both PWSMA and caregivers is the assumption on the part of education officials that a child with a physical disability should be placed in the special education class (i.e. due to learning disability), even though the risk of learning disabilities for PWSMA is the same as the general population.
>
> – *Living With Spinal Muscular Atrophy in Malaysia: A Study*

➢ Lack of adequate infrastructure

> From the point of view of the infrastructure, public facilities like public toilets are not disabled-friendly. The whole list can go on and on whichever place you go. And of course, the working place – if he starts working, we of course would expect that environment to be disabled-friendly.
>
> – *A father of a young man with SMA type 2*

BARRIERS TO SOCIAL INCLUSION

➢ Lack of awareness about SMA

> When they see us, they think we can do things by ourselves, they think that we are normal, but we really do not have much strength to do that – we need their assistance.
>
> – *A woman in her 40s with SMA type*

➢ Lack of adequate infrastructure

> In Malaysia, a lot of places are still not accessible to wheelchairs - this is one of the concerns. Even if we want to bring her to as many places as we wish, because wheelchair accessibility is very poor in Malaysia, sometimes we won't bring her there. We can't, so we don't go because of the inconvenience.
>
> – *A mother of a woman with SMA type 22*

➢ Lack of compassion

> A huge problem with the lifts ... if people are trying to get in the lift, even though they see there is a [person in a] wheelchair there, they don't wait for me [to go in]. So I just drag myself to the side, and I just let them all go in, and I will just wait for the next one ... The facilities are not maintained [and] the inconsideration of people parking in the disabled spot.
>
> – *A woman in her 20s with SMA type 30*

POLICY RECOMMENDATIONS

- ➢ Ensure that the principle of inclusive education is fully implemented in all schools.
- ➢ Enforce the Zero Reject Policy that guarantees every child in Malaysia access to education.
- ➢ Train all teachers in disability awareness and the basics of special education needs.
- ➢ Propose a law that prevents discrimination against persons with disabilities when it comes to employment.
- ➢ Reintroduce the proposed amendments to protect job seekers against discrimination to the Employment Act 1955, which were withdrawn in 2019.

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Our worries and wants

These are the worries and concerns, as well as the hopes and wants, of persons with spinal muscular atrophy (PWSMA) and their caregivers.

WORRIES AND CONCERNS

1. Losing their independence or never being able to be independent
2. Losing their functional abilities
3. Not being able to lead a normal life
4. Unable to join in social activities due to lack of accessibility
5. Lack of employment opportunities
6. The high cost of raising a child(ren) with SMA
7. Decreased mental and emotional health due to:
   - ➢ Loss of sleep and stress
   - ➢ Feelings of frustration and hopelessness
   - ➢ Feelings of uncertainty and anxiety
   - ➢ Depression and loneliness
   - ➢ Feelings of grief and sadness
8. Facing premature death, which includes:
   - ➢ The fear of dying or your child dying
   - ➢ Preparing to die
   - ➢ Living a meaningful life
9. Facing difficult treatment choices

HOPES AND WANTS

1. Access to disease-modifying treatments and clinical trials

2. Access to holistic post-diagnosis care, including:
   - ➢ Palliative care support
   - ➢ Professional mental health counselling
   - ➢ Respite care
   - ➢ A patient support group
   - ➢ Personal assistants for adult PWSMA

3. Help from the Government to:
   - ➢ Increase awareness about SMA
   - ➢ Improve accessibility for ease of movement for PWSMA, to education and employment, as well as to promote societal inclusivity for PWSMA
   - ➢ Improve medical care services for SMA, e.g. subsidise or reimburse treatment costs

People are not disabled by their medical condition, but rather the attitudes and structures of society, says the social model of disability. When these barriers are removed, disabled people can be independent and equal in society, with choice and control over their own lives.

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Treatments that matter

These are the treatments that persons with spinal muscular atrophy (PWSMAs) and their caregivers consider the most important to them.

While SMA has no cure, there are a variety of treatments that can **improve the quality of life** of the PWSMA. Of crucial importance is to **diagnose the condition** and **start treatment as soon as possible** in order to best manage the complications of the disease.

"Untreated SMA can have devastating consequences."

– *Living With Spinal Muscular Atrophy in Malaysia: A Study*

MOST IMPORTANT OUTCOMES FOR TREATMENT

- **To provide gains in function**
- **To lessen symptoms**
- **To stop or slow down disease progression**
- **To prolong lifespan***

**Note: Only caregivers chose this option*

"When asked to indicate what would be the most important outcome for a possible drug treatment, providing gains in function, such as increased strength and energy, and being able to do something the PWSMA was unable to do before, were the most important outcomes for both PWSMA and caregivers."

– *Living With Spinal Muscular Atrophy in Malaysia: A Study*

CRUCIAL TREATMENT AREAS

DISEASE-MODIFYING TREATMENT

There are currently three drugs that can treat SMA: nusinersen (Spinraza), onasemnogene abeparvovec-xioi (Zolgensma) and risdiplam (Evrysdi).*

➢ Respiratory care

"Lung disease is of primary concern as it is a main cause of death in SMA. The type of SMA will determine the extent of respiratory involvement. A proactive approach of introducing respiratory therapies early in the disease is the way forward in treating the pulmonary manifestations of SMA."

– *Living With Spinal Muscular Atrophy in Malaysia: A Study*

"Cough assist machine (CAM) has often enabled me to do my daily activities without being distracted by phlegm. Before I had started to use CAM, I was frequently struggling with phlegm in the morning as I always wake up with phlegm in lungs. ... Since I began to use CAM, I have been able to cough out my phlegm in the morning. It has assisted me to clear my lungs before I start the day. ... All in all, CAM has improved my lifestyle."

– *A man in his 20s with SMA type 2*

➢ Rehabilitation and orthopaedic care

"Rehabilitation goals differ according to the different types of SMA and the course of the disease, but these goals should also depend strongly on the patient's and family's wishes. ... Some of these goals include optimisation of function, minimisation of impairment and prevention of contractures and scoliosis."

– *Living With Spinal Muscular Atrophy in Malaysia: A Study*

➢ Palliative care

"I think palliative care is very, very important. The palliative team needs to come in and talk to the parents. Even if I know my child is going to pass on, what is the best I can do during this period? ... We have cases where parents who are given palliative consultation and all that, feel like, 'Okay, I have done my best, it's okay. That is the journey my child ... will finally go on.' If the parents are given that kind of counselling, besides genetic counselling, it gives a very good feeling, as if you are prepared, you have tried your best."

– *A mother of a boy with SMA Type 1*

FOR CAREGIVERS, RESPITE CARE IS ALSO CRUCIAL

"We are very tired! And if the parents or the caregivers are not taken care of, it doesn't only affect the child with SMA, but the whole family. ... Our hope is to have support for caregivers like us. The Government or other NGOs can come in with a system that actually supports caregivers, like what they do in Australia, where maybe once in a week or once in a month, someone comes in to help relieve the caregivers, which I think is very, very important."

– A mother of a boy with SMA Type 1

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Needs and recommendations for doctors

Doctors play a crucial role in these areas for persons with spinal muscular atrophy (PWSMA) and their caregivers.

"While SMA may be considered a rarity by most clinicians, new advances in SMA management, such as the emergence of novel therapies, has escalated the need for SMA education and awareness among all levels of healthcare providers, especially paediatricians and general practitioners."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

DURING DIAGNOSIS

➢ Developmental delays need to be recognized early.

"There is a lack of knowledge and very little attention or urgency given to developmental delay in infants among healthcare professionals, as evident from expressions such as 'It's nothing' and 'Biasalah (It's normal)'."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

➢ Sensitivity and compassion are crucial when giving the diagnosis.

"Doctors lack empathy and did not offer much help. The doctor said, 'Eh, you know or not, your child ah, can't live long. I can only give you a period of a maximum of two years.'"

– A father of two children with SMA Type 1

AFTER DIAGNOSIS

➢ Multidisciplinary care and collaboration are essential.
Among the main areas of concern for persons with SMA and their caregivers are:

➢ Help with referrals for financial aid and resources like SMA support groups.

➢ Listen to and work with PWSMA and their caregivers.

"Yes, we understand that the doctors cannot know everything, but try to work with the parents. Trust the parents or listen to what the parents want first. Don't see our child as a disorder, see our child as a human first."

– A mother of a boy with SMA Type 1

"Support groups can provide mutual aid and information- sharing, which enables parents to feel less helpless and more adequate in caring for their children."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

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Needs and recommendations for policymakers

Policymakers can play a crucial role in these areas for persons with spinal muscular atrophy (PWSMA) and their caregivers.

"Fundamentally, the government needs to endorse national plans for rare diseases that provide frameworks incorporating a coordinated 'whole-of-government' approach to rare diseases, as opposed to a piecemeal approach. These plans should outline a cohesive clinical, public health and disability service approach to rare diseases that addresses prevention, timely diagnosis, early intervention, and appropriate access to treatments and rehabilitation."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

➢ Improve accessibility and promote inclusivity of PWSMA.

This includes ensuring equal opportunities for education and employment.

"I think the number one thing would be to make places more accessible. ... I mean, they have no idea how important it is - a small thing such as a ramp can make such a big difference, you know."

– A man in his 20s with SMA type 2

"If you have a system that appreciates you as a human being, you won't be driven to the corner by people who don't know what or who you are. There will be no stigma because stigma has been eradicated by simple laws against discrimination."

– A woman in her 30s with SMA type 2

➢ Ease the process of hiring personal living assistants for PWSMA.

This includes reducing the cost of hiring foreign domestic helpers who serve in such capacities.

"This is my biggest concern, because if I can't find a helper, I would feel like I have lost both limbs and not be able to function."

– A woman in her 40s with SMA type 2

➢ Provide adequate resources for financial aid.

This includes providing an allocation in the annual Budget for PWSMA and outlawing genetic discrimination by insurance companies.

➢ Facilitate access to clinical trials and new drugs for SMA.

"Untreated SMA can have devastating consequences. Timely identification of symptoms and prompt referral to a specialised centre is critical to avoid a long diagnostic odyssey and enable earlier access to evidence-based care and intervention, as well as informing reproductive planning and reducing patient and parental stress."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

➢ Provide visibility and increase awareness of spinal muscular atrophy (SMA) at a national level.

This includes creating a national SMA registry.

"To them, some of us may look rather normal, but they don't know that we cannot even lift our hands."

– A woman in her 40s with SMA type 2

"The expense of purchasing assistive devices, mobility aids, respiratory support equipment (e.g. ventilators, BiPAP, CPAP and cough assist machine) and home modifications are exorbitant and are not covered by insurance, forcing some parents to find a second job, take out loans or look for charitable assistance."

– Living With Spinal Muscular Atrophy in Malaysia: A Study

"My suggestion and top priority for the Government is to bring in Spinraza*. And put aside a certain allocation, you know, every year for SMA patients."

– A father of a boy with SMA Type 3

**At the time of interview, only Spinraza was approved for the treatment of SMA. Currently, there are two other drugs also approved for SMA treatment: Zolgensma and Evrysdi.*

➢ Enable a national screening process for SMA carriers (adults with one mutated copy of the SMN1 gene) and fetuses with SMA via prenatal diagnosis, carrier screening and/or newborn screening.

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