

JR is a child diagnosed with SMA, he has lost his active mobility of all four limbs except below his right elbow. He is completely confined to a wheelchair. He has poor lung function and digestive function. His parents found out he was diagnosed with this disease when he was 5 months old, they were just shocked to observe something wrong related to his muscle strength. They couldn't initially accept the fact when his diagnosis was confirmed as incurable SMA. He has now been diagnosed with the disease for 14 years, they are not financially stable to pay for the treatment and need support. The doctors have helped them a lot so far by providing multi-disciplinary care which is essential for SMA Management.

Superheroes behind the action

Dr. Ilin Kinimi is a consultant and pediatric pulmonologist at Manipal Hospitals, Bangalore. She's done a fellowship in pediatric pulmonology and sleep medicine from NUH Singapore and diplomate ERS in pediatric respiratory medicine.

Dr. Ann Agnes Matthew completed her MBBS from St. John's Medical College, Bangalore, and passed out as the best outgoing student in 2000. She went on to do her Paediatric training in Scotland, United Kingdom.



What is SMA? - [Spinal Muscular Atrophy](#) is a genetically inherited disease that causes gradually worsening weakness due to damage to the anterior horn cells that are present in the spinal cord and control our muscles. It is the commonest cause of inherited infant deaths. It is a hereditary disease that destroys the nerves responsible for controlling voluntary muscle movement but does not affect intelligence. Muscles that control breathing, swallowing, head and neck control, walking, and crawling are the most severely affected. SMA is a variable disease in terms of when the symptoms begin. Most often it shows up before the child is 2 years old, but in some individuals, the symptoms can appear at a later age. Over 60% of individuals who are diagnosed with SMA are severely affected.

Most patients find it hard to sustain (financially) themselves during treatment-
“Most of the treatment and long-term management for these children require

multi-disciplinary teams and logistics support. For any child to benefit it requires a lot of financial decisions as well as psychosocial support –which is not possible by a single person. Hence for these patients to avail of all these – we decided to run this clinic at a subsidized cost whereas most Medical Doctors provide their service for free.,” Dr. Ilin Kinimi commented.

To help out patients and families affected by SMA, the best possible way is to spread awareness of the disease to the public.- “The best way of spreading awareness is by educating the general public as well and empowering families in taking care of affected children. We do this through awareness campaigns, parents and family days for SMA-affected individuals, and writing articles.

Young people could also help spread awareness by educating through blogs and other means of social media, in ensuring useful information such as the fact that it is best to avoid getting married among relations as that would lead to children being affected with genetic diseases like SMA to be disseminated.”, Dr. Ann said.



Ishals story -

Ishal Maryam is a patient diagnosed with SMA. SMA has affected her overall mobility and breathing, she was diagnosed with the disease when she was just one and half months old and has been diagnosed with the disease for the past year. Upon finding out about the diagnosis, her family was devastated, they always saw their kid growing up normally like any other child but the doctors have been supportive, they’ve given them hope and mental support and have taken care of all the problems so far. The family is financially unstable, but the doctors have given most of the treatment free of cost.

Effects - SMA symptoms cover a broad spectrum, ranging from mild to severe. The muscles most affected are those closest to the center of the body, such as

those of the shoulders, hips, thighs, and upper back. In many forms of SMA,



weakened respiratory muscles make it difficult to cough and clear secretions, leading to an increased risk of serious respiratory infection. A simple cold can quickly progress to pneumonia. If the muscles of the back weaken, spinal curvatures can develop. SMA does not affect emotional development, learning

or academic ability, or sensory ability. It makes affected children gradually lose control over their body and so doing things like walking, sitting, or even having head control, breathing, and swallowing becomes a challenge, and ultimately that is how the disease becomes life-threatening. One is unable to breathe well, cough effectively, or swallow safely.

There's still hope.....

Spinraza: In December 2016, [FDA](#) approved the treatment of all types of SMA in children and adults. Spinraza is administered by intrathecal injection into the fluid surrounding the spinal cord and is designed to increase the production of full-length SMN protein.

Zolgensma: In May 2019, [FDA](#) approved the treatment of children less than 2 years of age. Zolgensma is a gene therapy administered by a one-time intravenous injection. This therapy results in the long-term production of full-length SMN protein within motor neurons, improving muscle function and survival.

Risdiplam: in August 2020, [FDA](#) approved the treatment of SMA in adults and children two months of age or older. It is an oral medication designed to increase levels of the SMN protein by enhancing production from the SMN2 “backup” gene.



[ORDI- Organisation for Rare diseases India](#) is a national umbrella organization representing the collective voice of all patients with rare diseases in India, set up as a section 25 non-profit company in India. ORDI has helped SMA patients all over India raise funds and spread awareness about the disease.

[Impact Guru](#) is one of the biggest crowdfunding platforms in India, it helps to raise funds for those who cannot afford medical treatments and long-term care. It has helped a lot of SMA patients raise funds for their treatment.

This disease is affecting more and more children day by day, but doctors are also working together to find cures and rid children of this disease. many doctors across the world have taken the initiative to run free clinics and provide free treatment to SMA patients.