ORGANIZATION FOR RARE DISEASES INDIA

Special Recognition for Rare Star Awards

We are profoundly grateful to the exceptional individuals and organizations whose unwavering dedication, pioneering efforts, and extraordinary contributions have significantly advanced the cause of rare diseases. Their commitment to innovation, advocacy, and support has had a transformative impact on countless lives. We thank them for being beacons of hope and progress in the rare diseases community.

Dr. Anne Agnes

Dr. Meenakshi Bhatt

Team Sanofi

Team IQVIA

Dr. Sanjeeva

Our Heartfelt Appreciation for the Jury Members of Rare Star Awards

We extend our heartfelt gratitude to the esteemed jury members of the Rare Star Awards for their invaluable contribution in celebrating the 10th anniversary of ORDI and DNA Day. Your commitment to excellence and fairness has ensured that the most deserving individuals and organizations are acknowledged for their remarkable efforts in advancing the cause of rare diseases. Thank you for your unwavering support and for making this milestone celebration a resounding success.

Genetic Counselor Category

Researcher & Scientist category & Health Innovation Startup Category

Advocacy Leader category, Caregiver category & Role Model Living with Rare Category

PAG Interview: Scleroderma India

This month, we had the privilege of speaking with Rashmi Bhasin, a Trustee and Core Committee member of Scleroderma India. Rashmi is not only a dedicated advocate for scleroderma awareness but also a warrior who has been battling this challenging condition for over a decade. Her personal experience and commitment to the cause provide valuable insights into the struggles faced by scleroderma patients and the crucial work being done to support them. Through this conversation, Rashmi sheds light on the foundation's origins, the obstacles in diagnosing and managing the disease, and her own journey of resilience and hope.

1. Can you share the story behind the foundation of Scleroderma India?

Scleroderma India was founded by Neetu Wadhwa, who sadly is no longer with us. Neetu began experiencing health issues in 2007, which went undiagnosed until 2012 when she was finally diagnosed with scleroderma. By then, it was quite late, and her condition had severely deteriorated. Neetu founded the organization in 2017, driven by the belief that every patient deserves dignity.

One of the biggest challenges is the lack of understanding and awareness about this disease. Often, it's misdiagnosed as a mental illness, particularly because it predominantly affects women of reproductive age. Many patients, including myself, have been dismissed by doctors who suggest psychiatric help when they cannot diagnose the physical symptoms. The initial symptoms often affect the skin, leading patients to visit dermatologists who may provide incorrect treatments for years.

The foundation aims to spread awareness about scleroderma and educate patients on the importance of proper disease management. While doctors may prescribe numerous medications, they often neglect to discuss the critical aspects of managing the disease, such as exercise, diet, and sleep. We focus on these areas to improve the quality of life for patients.

2. What exactly is scleroderma?

Scleroderma is a chronic autoimmune disorder where the immune system attacks the body's own tissues, leading to excessive collagen production. This results in the hardening and tightening of the skin and connective tissues. Symptoms vary but commonly include skin thickening, color changes, Raynaud's phenomenon, joint pain, muscle weakness, and digestive issues.

Diagnosing scleroderma is challenging due to its overlapping symptoms with other disorders and often requires a combination of clinical evaluation and tests. While there is no cure, the disease can be managed with medications, lifestyle changes, and supportive therapies to improve the quality of life for patients.

3. Does scleroderma have a genetic basis?

No, the genetic basis of scleroderma has not been proven. We have yet to see multiple cases within a single family.

4. Does scleroderma have a genetic basis?

Unfortunately, the exact cause of scleroderma is unknown. This applies to many autoimmune disorders. The body's immune system attacks its own cells. We cannot pinpoint specific lifestyle factors or triggers that increase susceptibility to the disease.

5. Since the diagnosis of scleroderma is quite difficult, how do you assist patients in finding the right doctors and getting proper treatment?

Many patients are initially misdirected to dermatologists due to skin-related symptoms. However, scleroderma is best treated by rheumatologists who specialize in autoimmune conditions. The disease causes the immune system to attack the body, and its symptoms overlap with many other disorders, making diagnosis challenging.

Doctors often fail to dig deep enough to diagnose scleroderma correctly. Their communication can also be problematic; telling patients that there is no cure can be very discouraging. It's crucial to convey that, while scleroderma cannot be cured, it can be managed effectively with the right treatments, much like diabetes.

6. What are the daily challenges that patients face when living with scleroderma?

Patients face numerous daily challenges. Scleroderma is an invisible disability; outwardly, patients may appear fine, leading to misunderstanding and mistreatment. This is a common issue with many chronic conditions. At home, conceiving can be difficult due to medications, leading to societal stigma. Some days are good, others are bad, and people often misunderstand this inconsistency, accusing patients of making excuses. Simple tasks like washing dishes can be painful due to the condition affecting the skin and causing ulcers.

The visible symptoms, such as skin tightening and color changes, lead to social stigma. People may avoid you, assuming the condition is contagious. Physical changes, like claw-like fingers and stooped posture, further isolate patients socially and professionally. Employers often fail to accommodate the needs of patients, leading to job loss despite their ability to perform intellectually.

Scleroderma is also an expensive condition. Monthly expenses for tests, doctor visits, and medications can range from 25,000 to 30,000 rupees for mild cases and up to 50,000 to 60,000 rupees for more severe cases. Despite being a rheumatic condition, it is often not covered by insurance. Our foundation advocates for patients, ensuring they are not overcharged for unnecessary treatments and medications, and provides support systems to help them navigate these challenges.

7. What are the earliest signs of scleroderma that people should look out for?

The two major red flags are skin changes—tightening, shiny appearance, and color changes—and a persistent cough that doesn't go away. While these symptoms are common and may not seem serious, they can indicate something more significant.
Other signs include difficulty with daily tasks like holding a knife or wearing clothes due to pain and stiffness. It's important not to panic but to consult a rheumatologist if these symptoms persist and seek proper diagnosis and management.

8. What message would you like to give to patients recently diagnosed with scleroderma and their families?

Don't let scleroderma define your life. It's possible to live a normal life with proper management. I'm currently in remission and engage in activities like Pilates and swimming. It's important to incorporate the disease into your life without letting it take over. With the right treatment and management, scleroderma can be a small part of your life, not its focus.

Rashmi Bhasin's insights and experiences shed light on the profound challenges faced by those living with scleroderma. Her dedication to raising awareness and advocating for better care through Scleroderma India is truly inspiring. As a scleroderma warrior herself, Rashmi exemplifies resilience and hope, showing that with the right support and management, it is possible to live a fulfilling life despite the condition. We hope this interview not only educates but also encourages greater understanding and compassion for those affected by scleroderma. Together, we can work towards a future where no one has to face this battle alone.

WORLD THALASSEMIA DAY

On **May 8**, the global community observes World Thalassemia Day, a vital occasion dedicated to raising awareness about Thalassemia, a group of inherited blood disorders. This day is essential for educating the public, advocating for patients, and supporting research efforts aimed at better understanding and managing this condition.

What is Thalassemia?

Thalassemia is a genetic blood disorder affecting the production of hemoglobin, the protein in red blood cells responsible for carrying oxygen. The disorder can cause mild to severe anemia, depending on the type of Thalassemia. The primary forms include:

·**Alpha Thalassemia**: Alpha Thalassemia results from mutations in the HBA1 and HBA2 genes, which affect the production of alpha globin chains. There are four genes responsible for producing alpha globin chains, and the severity of Alpha Thalassemia depends on how many of these genes are mutated.

·**Beta Thalassemia**: Resulting from mutations in the HBB gene, ranging from mild (Thalassemia minor) to severe (Thalassemia major or Cooley's anemia).

Recognizing the Symptoms

The symptoms of Thalassemia vary in type and severity. Common signs include:

·**Chronic Anemia**: Leading to fatigue, weakness, and pale or jaundiced skin.

·**Skeletal Deformities**: Particularly in the face and skull.

·**Growth Retardation**: Delayed growth and development in children.

·**Splenomegaly**: Enlargement of the spleen due to excessive red blood cell breakdown.

·**Iron Overload**: Frequent blood transfusions can cause iron accumulation, damaging organs such as the heart and liver.

World Thalassemia Day emphasizes the importance of awareness, which can lead to earlier diagnosis, improved treatment options, and a better quality of life for those affected. Early detection and proper management can significantly reduce the risk of complications for many patients.

WORLD LUPUS DAY

World Lupus Day, celebrated annually on **May 10**, is a crucial event dedicated to increasing awareness about lupus, an often misunderstood autoimmune disease. This day provides an opportunity to educate the public, advocate for patients, and support ongoing research efforts aimed at finding better treatments and a potential cure for lupus.

What is Lupus?

Lupus is a chronic autoimmune disease where the body's immune system attacks its own tissues and organs. The inflammation caused by lupus can affect various systems, including the joints, skin, kidneys, blood cells, brain, heart, and lungs. Lupus is characterized by periods of illness, called flares, and periods of remission when symptoms improve.

The Many Faces of Lupus

Lupus is a complex disease with different manifestations. The main types include:

·**Systemic Lupus Erythematosus (SLE)**: The most common form, affecting multiple organs and systems.

·**Cutaneous Lupus**: Limited to the skin, causing rashes and lesions.

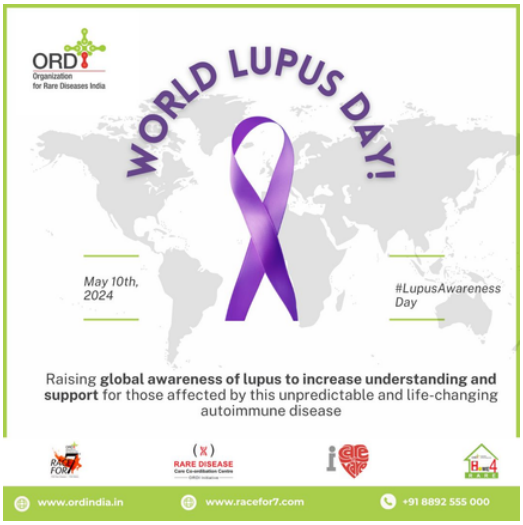
·**Drug-Induced Lupus**: Triggered by certain medications, with symptoms usually subsiding once the medication is stopped.

·**Neonatal Lupus**: A rare condition affecting newborns of women with lupus, causing skin rash, liver problems, and low blood cell counts.

Symptoms:

Lupus symptoms vary widely among individuals, often mimicking other illnesses, which makes diagnosis challenging. Common symptoms include fatigue, joint pain and swelling, particularly in the hands, wrists, and knees, skin rashes (notably the butterfly-shaped rash across the cheeks and nose), fever, inflammation of organs.

Diagnosis typically involves a combination of laboratory tests, symptom assessment, and medical history review. Tests may include blood and urine tests, imaging studies, and biopsies of affected tissues.



The Importance of World Lupus Day

World Lupus Day is pivotal in shining a light on lupus, a disease that often remains in the shadows. Increased awareness can lead to earlier diagnosis, more effective treatments, and better support for those affected. Public understanding of lupus can also drive funding for research and advocacy efforts

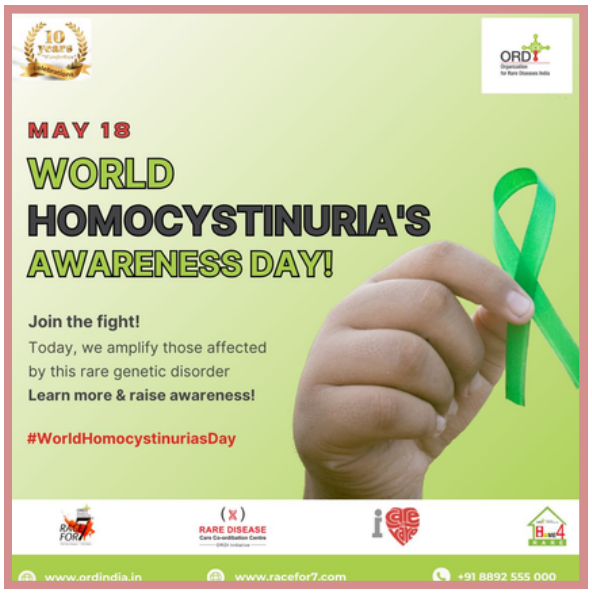
World Lupus Day is a significant occasion that underscores the importance of awareness and community support in the fight against lupus. By coming together on May 10, we can make a meaningful impact on the lives of those affected by lupus, offering hope and fostering progress towards better treatments and, ultimately, a cure. Let us stand in solidarity with lupus patients, amplifying their voices and championing their cause.

WORLD HOMOCYSTINURIA DAY

Every year on **May 18**, the global community comes together to observe World Homocystinuria Day, a crucial day dedicated to raising awareness about Homocystinuria, a rare genetic disorder. This day serves as a platform to educate the public, advocate for patients, and support research efforts aimed at better understanding and managing the condition.

What is Homocystinuria?

Homocystinuria is a metabolic disorder characterized by an inability to properly process certain amino acids, specifically methionine. This leads to an accumulation of homocysteine in the blood and urine, which can cause a variety of health issues, including developmental delays, eye problems, skeletal abnormalities, and vascular complications.



The condition is typically inherited in an autosomal recessive pattern, meaning that a child must inherit a defective gene from both parents to develop the disorder. While it is a rare disease, affecting approximately 1 in 200,000 to 1 in 300,000 individuals worldwide, its impact on patients and families is profound.

Diagnosis and Treatment

Early diagnosis of Homocystinuria is essential for effective management. Newborn screening programs can detect the disorder soon after birth. Diagnostic tests include blood and urine analysis to measure homocysteine levels and genetic testing to identify mutations.

Treatment typically involves a combination of dietary modifications, supplements, and medications:

- **Dietary Changes**: Low-protein diets and special formulas to reduce methionine intake.
- **Supplements**: Vitamin B6, B12, and folic acid to help lower homocysteine levels.
- **Medications**: Betaine and other drugs to manage symptoms and prevent complications.

World Homocystinuria Day is a reminder of the power of awareness and community action. By coming together on May 18, we can make a significant difference in the lives of those affected by Homocystinuria, fostering hope and progress in the journey towards a better understanding and management of this rare disease. Let us stand in solidarity with patients and their families, amplifying their voices and championing their cause.

Neurofibromatosis Awareness Day: Educate, Support, Advocate

Neurofibromatosis Awareness Day is an important event dedicated to increasing public understanding and support for individuals affected by neurofibromatosis (NF). Observed annually on May 17th, the day serves as a focal point for education, advocacy, and fundraising efforts aimed at combating this genetic disorder.

1. **Neurofibromatosis Type 1 (NF1):** The most common form, NF1 is characterized by multiple café-au-lait spots on the skin, neurofibromas (benign skin tumors), and an increased risk of learning disabilities. It can also lead to more serious complications such as scoliosis, vision problems, and an increased risk of certain cancers.

2. **Neurofibromatosis Type 2 (NF2):** Less common than NF1, NF2 primarily affects the development of benign tumors on the nerves responsible for hearing and balance, leading to hearing loss, tinnitus, and balance issues. These tumors, known as vestibular schwannomas or acoustic neuromas, can cause significant complications if left untreated.

3. **Schwannomatosis**: The rarest form of NF, schwannomatosis causes the development of multiple benign tumors called schwannomas, typically on spinal and peripheral nerves. Unlike NF2, it does not usually involve tumors on the auditory nerves, and hearing loss is not a common symptom. However, schwannomatosis can cause chronic pain and neurological dysfunction.

THE IMPORTANCE OF AWARENESS

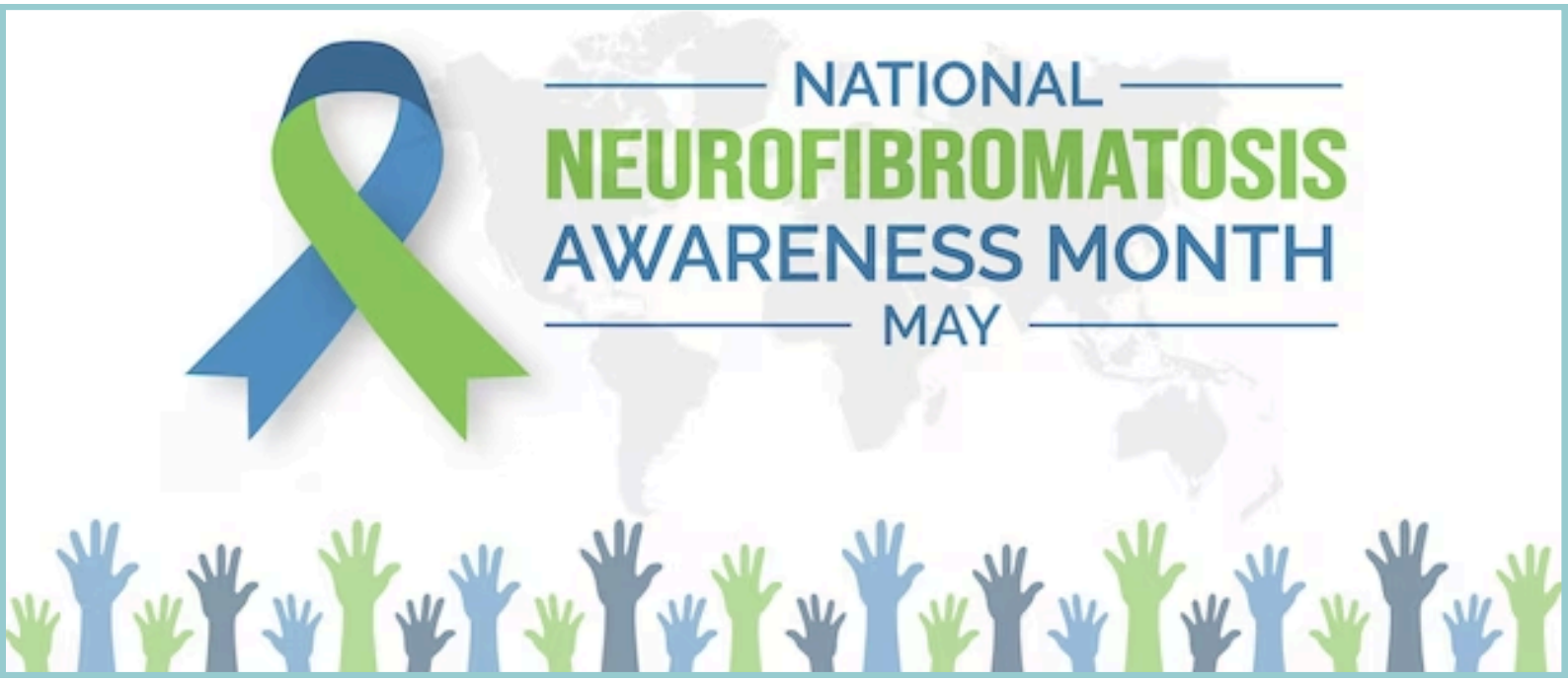
Raising awareness about neurofibromatosis is crucial for several reasons:
1. **Early Diagnosis and Treatment:** Increased awareness can lead to earlier diagnosis and intervention, improving the quality of life for those affected. Early detection allows for better management of symptoms and prevention of severe complications.

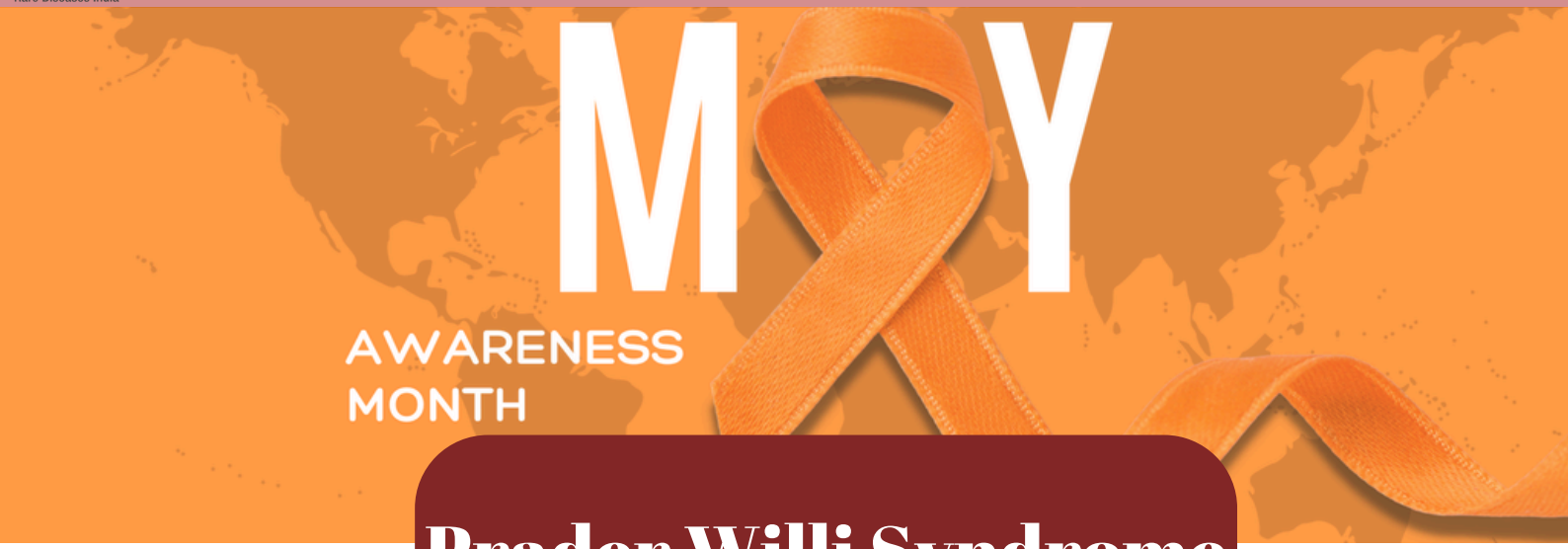
2. **Research and Funding:** Public awareness campaigns help garner support and funding for research into the causes, treatments, and potential cures for neurofibromatosis. This funding is vital for advancing medical knowledge and developing new therapies.

3. **Support and Community**: Awareness days foster a sense of community among individuals and families affected by NF, providing them with opportunities to connect, share experiences, and support each other.

4. **Reducing Stigma:** Educating the public about NF helps reduce the stigma and misconceptions associated with the disorder. This can lead to greater empathy and inclusion for those living with NF in schools, workplaces, and social settings.

Neurofibromatosis Awareness Day plays a crucial role in shining a light on this often-overlooked condition. Through collective efforts in education, fundraising, and advocacy, we can make significant strides toward better understanding, treating, and ultimately curing neurofibromatosis





Prader Willi Syndrome

Understanding Prader-Willi Syndrome

Prader-Willi Syndrome (PWS) is a complex genetic disorder resulting from the loss of function of specific genes on chromosome 15. Key characteristics include:

- **Hypotonia:** Poor muscle tone, particularly noticeable in infants.
- **Hyperphagia:** An insatiable appetite leading to chronic overeating and obesity if not controlled.
- **Growth Hormone Deficiency:** Resulting in short stature and incomplete sexual development.
- **Cognitive and Behavioral Challenges:** Learning disabilities, temper outbursts, and obsessive-compulsive behaviors.

The Importance of Awareness

Raising awareness about PWS can lead to earlier diagnosis and better management of symptoms, improving the quality of life for those affected. Public education helps reduce stigma and fosters understanding of the challenges faced by individuals with PWS and their families.

Tuberous Sclerosis Complex

Tuberous Sclerosis Complex (TSC) is a genetic disorder that causes benign tumors to develop in various organs, especially the brain, heart, kidneys, lungs, and skin. It is caused by mutations in either the TSC1 or TSC2 genes.

Neurological Symptoms: Seizures, developmental delays, and behavioral issues such as autism spectrum disorder.

Skin Abnormalities: Light-colored skin patches, facial angiofibromas, and shagreen patches.

Organ Complications: Kidney problems, heart tumors, and lung issues.

The Importance of Awareness

Awareness of Tuberous Sclerosis Complex (TSC) is vital as it facilitates early diagnosis and intervention, which can significantly improve the quality of life for those affected. Early recognition of TSC symptoms enables timely management of seizures, developmental delays, and other related complications, reducing long-term impacts on health and development. Additionally, heightened awareness fuels advocacy efforts that are crucial in securing research funding. This funding is essential for developing new treatments and potential cures, thereby offering hope for more effective management strategies and improved outcomes for individuals with TSC.

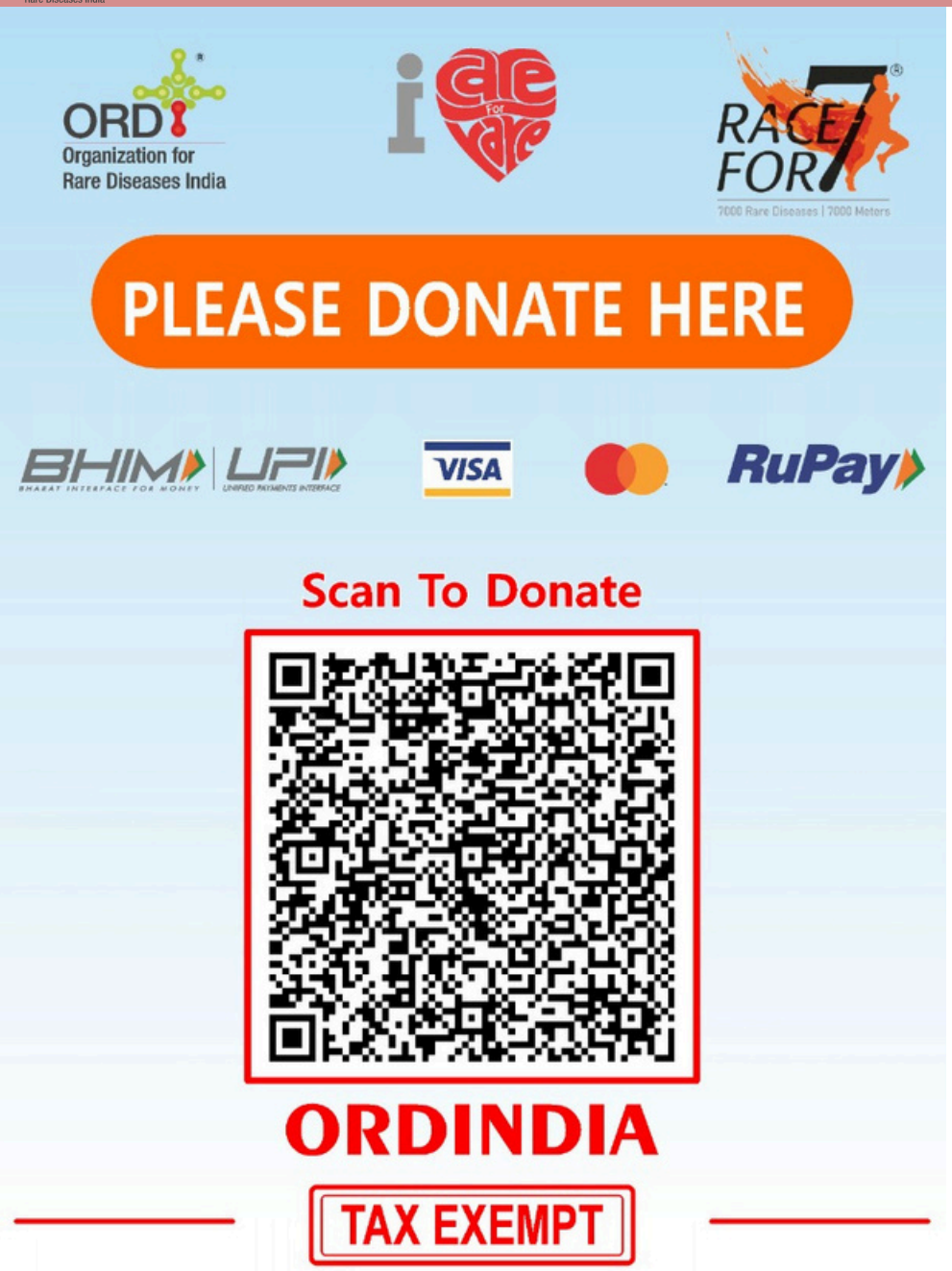
MUCOPOLYSACCHARIDOSES

Mucopolysaccharidoses (MPS) are a group of metabolic disorders caused by the absence or malfunctioning of lysosomal enzymes needed to break down glycosaminoglycans (GAGs). These disorders lead to a buildup of GAGs in cells, causing progressive damage to various tissues and organs. Common symptoms include:

- **Skeletal Abnormalities:** Joint stiffness and abnormal bone growth.
- **Organ Enlargement:** Hepatomegaly and splenomegaly.
- **Cognitive Impairment:** Developmental delays and intellectual disability in some types.
- **Respiratory Issues**: Obstructive airway disease and recurrent infections.

The Importance of Awareness

Raising awareness of MPS is essential for early diagnosis and treatment, which can improve the quality of life. Enzyme replacement therapy (ERT) and other treatments can help manage symptoms and slow disease progression. Public knowledge and support drive research efforts towards better treatments and potential cures.



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