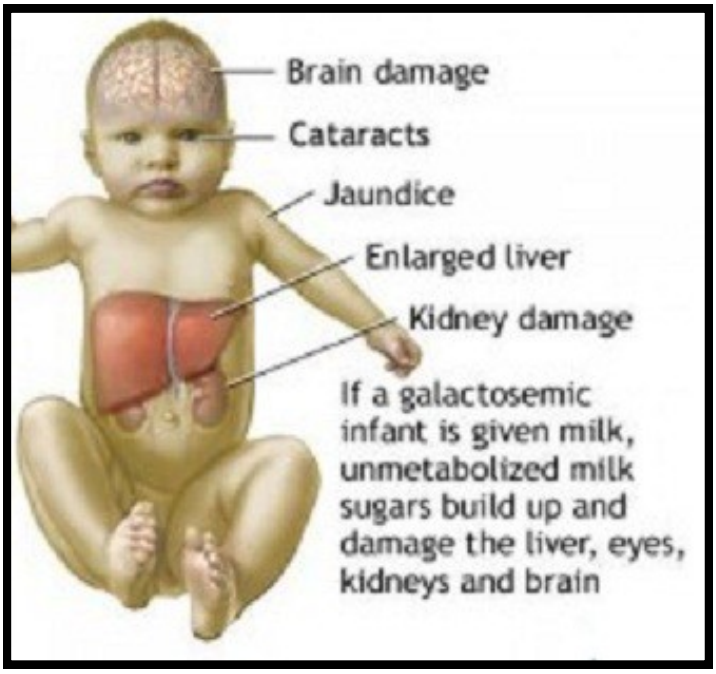
ORGANIZATION FOR RARE DISEASES INDIA

Inborn Errors of Metabolism

Unveiling Genetic Disorders for Better Health

Inborn errors of metabolism are a diverse group of disorders that can be inherited or arise from spontaneous mutations. These diseases result from failures in metabolic pathways responsible for breaking down or storing carbohydrates, fatty acids, and proteins. Although each specific inborn error of metabolism is rare, collectively they are quite common, occurring in about 1 in 2,500 births. They can manifest at any age, making it essential for emergency providers to have a comprehensive understanding of these diseases, their symptoms, and appropriate evaluation methods.

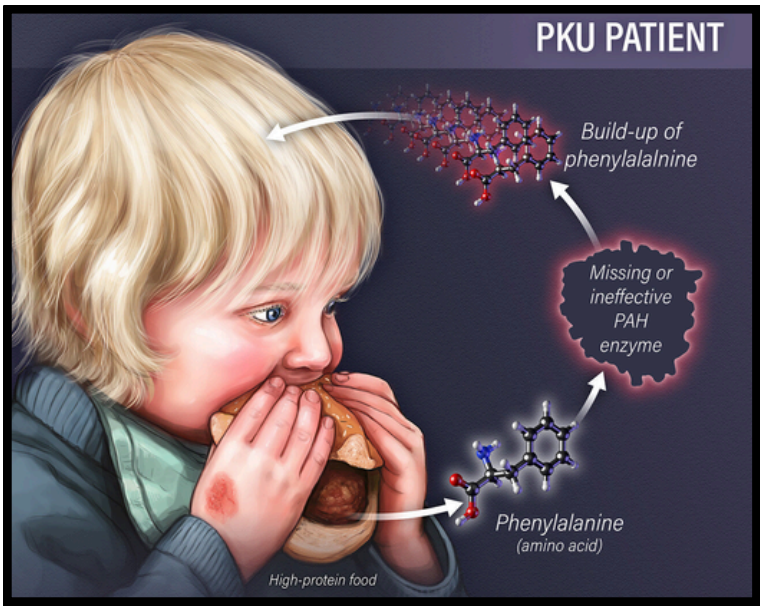
Maple Syrup Urine Disease



Maple Syrup Urine Disease (MSUD) is a rare, inherited metabolic disorder that affects the body's ability to break down certain amino acids found in protein. It is named for the distinctive sweet-smelling urine of affected individuals, which resembles maple syrup. MSUD is caused by mutations in the genes that encode the enzyme complex responsible for breaking down the branched-chain amino acids: leucine, isoleucine, and valine. The buildup of these amino acids and their toxic by-products can lead to severe health problems.

Phenylketonuria

Phenylketonuria (PKU) is a genetic disorder that results in the inability to metabolize phenylalanine, an amino acid found in many protein-containing foods. If untreated, phenylalanine can accumulate in the blood and brain, leading to intellectual disability and other serious health problems.PKU is caused by mutations in the PAH gene, which encodes the enzyme phenylalanine hydroxylase. This enzyme is necessary for converting phenylalanine into tyrosine, another amino acid. Without this enzyme, phenylalanine builds up in the body.



Pioneering Hope: Kamal Shah's Journey to Revolutionize Dialysis Care in India

Interview by Vaidehi kadam

Dialysis is critical for kidney failure, yet conventional methods pose many challenges. Kamal Shah, a long-term dialysis patient, has pioneered advancements in dialysis care in India, transitioning from limited dialysis to nocturnal home hemodialysis. In this interview, Kamal shares his experiences, obstacles faced, and vision for dialysis care in India. A role model within both the dialysis and atypical HUS international communities, Kamal exemplifies strength through overcoming challenges and continues to advocate for aHUS patients in India and beyond with his collaborative approach to optimizing information and opportunities through global action. Kamal D Shah is recognized as a key leader among the international aHUS community for his innovative solutions and team approach, and Atypical HUS India is a valued partner in the aHUS Alliance of global advocacy and patient organizations.

What is aHUS?

Atypical Hemolytic Uremic Syndrome (aHUS) is a rare and life-threatening disorder characterized by the formation of tiny blood clots in small blood vessels, leading to organ damage, particularly in the kidneys. It arises from uncontrolled activation of the complement system, often due to genetic mutations or acquired factors. Symptoms include fatigue, shortness of breath, bruising, swelling, and decreased urine output. Diagnosis involves blood tests, complement activity assays, and genetic testing to exclude other conditions. Treatment focuses on plasma exchange, complement inhibitors like Eculizumab, and supportive care. Early intervention is crucial for preventing long-term complications and improving patient outcomes.

The Atypical HUS India Foundation is a trust of patients, families, and doctors in India providing support and information about Atypical Hemolytic Uremic Syndrome (aHUS). It uses its website and social media to share details on the disease, testing, and treatments, bridging the gap between the Indian and global aHUS communities. As a member of the aHUS Global Alliance and the aHUS Community Advisory Board, it collaborates with pharmaceutical companies on drug development. Recently, the aHUS India Foundation has joined the Organization for Rare Diseases India (ORDI), expanding its network and support capabilities

Were there any initial challenges that you faced during treatment of aHUS and nocturnal home dialysis?

Yes, quite a few challenges arose initially. One major issue was finding a technician willing to stay overnight five to six nights a week to assist with the dialysis in case of complications. Additionally, there were significant costs involved in purchasing the dialysis machine setting it up at home, and ensuring a steady supply of consumables, which required coordination with suppliers. Fortunately, I received help from my technician and nephrologist, who assisted me in setting up the equipment and the supply chain.

What is your vision for the future of dialysis care in India?

While a diagnosis of kidney failure and the prospect of long-term dialysis can be daunting, it is possible to lead a completely normal life on dialysis. I have been on dialysis for 27 years and live a full life, swimming every morning, working full-time, traveling, and enjoying activities like going to movies and dining out. The key is to maintain a positive mindset and determination to live a normal life. It's important to fight for your life and keep a positive attitude.

How did NephroPlus address issues faced by dialysis patients in India?

We noticed that hospitals and doctors often viewed dialysis from their own perspectives, not the patients'. Our approach focused on the patient's experience from the moment they walked into the center until they left. We ensured the highest quality dialysis through strict protocols, ensuring the best possible treatment and clearance during sessions. This approach improved patients' quality of life, making them happier and more productive. Additionally, we introduced initiatives like holiday dialysis and the Dialysis Olympiad to promote normalcy in patients' lives, allowing them to travel and engage in sports activities.

How do you support the emotional and mental well-being of patients?

Our NGO, Aashayein Kidney Foundation, subsidizes treatment for those who can't afford it, helping with the cost of dialysis. Donations are tax-deductible under section 80G. People can also participate in organizing events like Aashayein and the Dialysis Olympiad, providing a gratifying experience by helping patients enjoy and learn more about their condition. These events are heartwarming and fulfilling, offering multiple ways to contribute to the community's effort.

Kamal Shah's journey from a patient facing the challenges of conventional dialysis to becoming a pioneer in nocturnal home hemodialysis and a leader in the aHUS community is truly inspiring. His efforts in organizing events like the Indian Dialysis Olympiad and advocating for better care and support for dialysis and aHUS patients demonstrate his dedication and resilience. Kamal's innovative approaches and collaborative spirit continue to offer hope and improve the quality of life for patients in India and around the world. His leadership within Atypical HUS India and the global aHUS Alliance underscores his commitment to making a significant impact on the lives of those affected by these conditions.

Facioscapulohumeral Muscular Dystrophy Awareness Day

LIVING WITH FSHD

Living with FSHD poses unique challenges. The progressive nature of the disorder means that muscle weakness tends to worsen over time, often leading to difficulties with daily activities such as lifting objects, reaching overhead, and even smiling. For many individuals, these physical limitations can also affect their emotional and psychological well-being, making comprehensive support systems and awareness crucial.

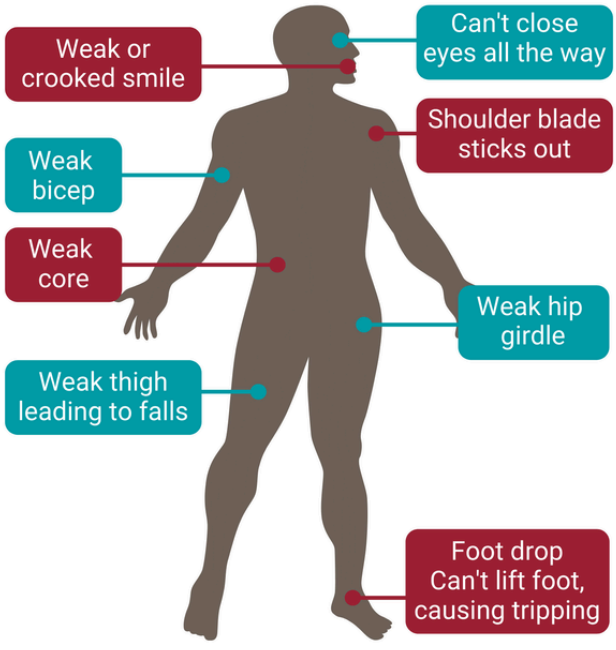
WORLD FSHD DAY

Unite to find a cure

Every year on June 20th, the global community comes together to raise awareness for Facioscapulohumeral Muscular Dystrophy (FSHD), a rare and often misunderstood condition. FSHD is a genetic muscle disorder that primarily affects the muscles of the face, shoulder blades, and upper arms. The condition derives its name from the specific areas it impacts: "facio" refers to the face, "scapulo" to the shoulder blades, and "humeral" to the upper arms. FSHD is characterized by progressive muscle weakness and atrophy, which can lead to significant physical limitations and a reduced quality of life.

This day serves as a crucial reminder of the importance of understanding, research, and support for those affected by FSHD. At ORDI, we are committed to spreading knowledge and fostering a supportive environment for individuals and families impacted by this challenging disorder.

About 1 in 8,000 people have FSHD.
That's nearly one million people worldwide.



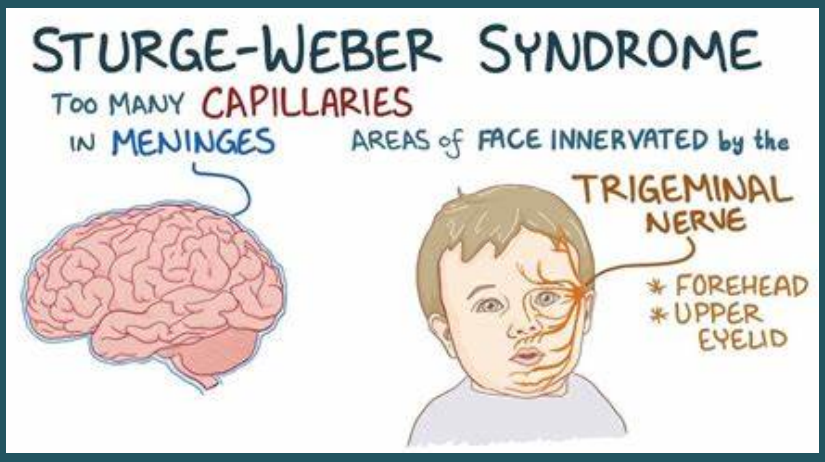
Sturge-Weber Syndrome Awareness Day - 27th June

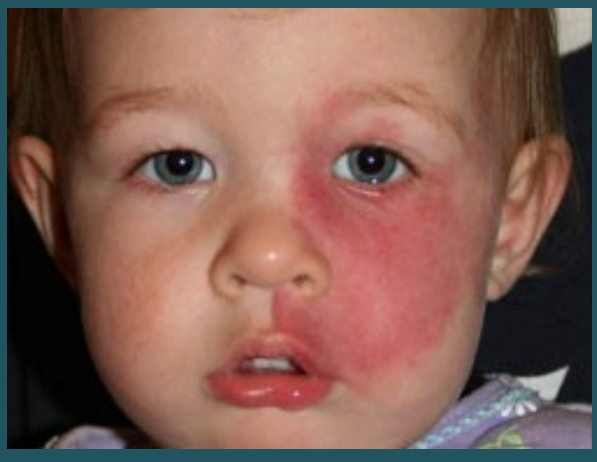
Sturge-Weber Syndrome (SWS) is a congenital disorder that is present at birth. It is characterized by a distinctive port-wine stain birthmark on the face, neurological abnormalities, and eye problems. The condition arises due to the abnormal development of blood vessels, specifically affecting the skin, brain, and eyes. The port-wine stain, a hallmark of SWS, is caused by an overabundance of capillaries near the surface of the skin.

The Challenges of Living with SWS

The impact of SWS extends beyond its physical manifestations. Individuals with SWS may experience seizures, developmental delays, and glaucoma. The neurological symptoms are particularly challenging, as they can lead to a range of complications, including hemiparesis (weakness on one side of the body), migraines, and intellectual disabilities. The visible nature of the port-wine stain can also lead to social and psychological challenges. Individuals with SWS often face stigma and misunderstanding from others, which can affect their self-esteem and mental health. The journey of living with SWS is one of resilience and courage, as individuals and their families navigate the complexities of this condition.

At ORDI, we are deeply committed to supporting the SWS community through education, advocacy, and research. In June and beyond, let us commit to making a positive impact in the lives of individuals with SWS. Together, we can build a more inclusive and supportive community for all.





International Batten Disease Day

June 9th marks International Batten Disease Day, a global initiative to raise awareness and support for those affected by this rare and devastating group of neurodegenerative disorders. ORDI stands in solidarity with the Batten disease community, advocating for greater understanding, research, and compassion.

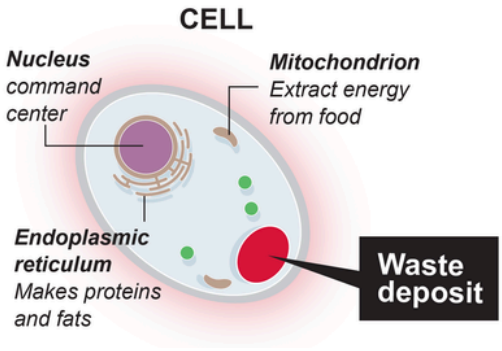
What is Batten Disease?

Batten disease, also known as neuronal ceroid lipofuscinosis (NCL), is a group of inherited disorders that primarily affect the nervous system. It is caused by mutations in genes responsible for producing proteins that help cells break down and dispose of waste. Without these proteins, waste accumulates in cells, leading to progressive neurological damage. Symptoms typically begin in childhood and include vision loss, seizures, cognitive decline, and motor impairment.

Human Impact

The diagnosis of Batten disease is a life-altering event for families. The progressive nature of the disorder means that children gradually lose their ability to see, move, and communicate, placing immense emotional and physical burdens on their families. Despite these challenges, the resilience and strength of these families are nothing short of inspiring.

◆ Fats and proteins called **lipofuscins** build up in cells of the brain, eyes and other tissues, creating deposits of waste.



Community & Support

One of the most powerful aspects of International Batten Disease Day is the sense of community it fosters. Families, healthcare professionals, researchers, and advocates come together to share their experiences, support one another, and work towards common goals. This collective effort creates a network of care that extends across borders, providing much-needed comfort and solidarity. Join us in spreading awareness and supporting the fight against Batten disease. Together, we can make a difference.

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