

Call for increased awareness on Acid Sphingomyelinase Deficiency as rare disease

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19th October marks the first Acid Sphingomyelinase Deficiency (ASMD) Awareness Day in India



Patients diagnosed with Acid Sphingomyelinase Deficiency (ASMD) and their caregivers have sought inclusion of this rare, genetic condition under the National Policy for Rare Diseases 2023, explaining that it will help create awareness and provide the much-needed support towards diagnosis, treatment.

Also referred to as Niemann-Pick Disease, ASMD is a rare progressive genetic disorder that results from a deficiency of the enzyme acid sphingomyelinase, essential for breaking down sphingomyelin - a fatty substance crucial for normal cellular function. ASMD is highly variable and the age of onset, specific symptoms and severity of the disorder can vary dramatically from one person to another, sometimes even among members of the same family.

The disorder may be best thought of as a spectrum of disease. At the severe end of the spectrum is a fatal neurodegenerative disorder that presents in infancy (Niemann-Pick disease type A). At the mild end of the spectrum, affected individuals have no or only minimal neurological symptoms and survival into adulthood is common (Niemann-Pick disease type B). Intermediate forms of the disorder exist as well. ASMD is caused by mutations in the SMPD1 gene and is inherited in an autosomal recessive manner.

Anil Raina, General Manager, Sanofi Speciality Care (India) said, "Sanofi has been at the forefront of launching therapies for rare diseases over the past 40 years, including the recent addition of Xenpozyme for the treatment of Acid Sphingomyelinase deficiency (ASMD). On the commemoration of the first ASMD Awareness Day in India, we join the global community in raising awareness about this debilitating disease."