GM2-Gangliosidosis, AB Variant

National Cancer Institute

Source

National Cancer Institute. GM2-Gangliosidosis, AB Variant. NCI Thesaurus. Code C133084.

An autosomal recessive lysosomal storage disease caused by mutation(s) in the GM2A gene, encoding ganglioside GM2 activator. It is characterized by GM2-ganglioside accumulation in tissues resulting in hypotonia, cherry-red macular spots, and neurocognitive dysfunction.