

Open Peer Review on Qeios

Alpha-Mannosidosis

National Cancer Institute

Source

National Cancer Institute. <u>Alpha-Mannosidosis</u>. NCI Thesaurus. Code C84548.

An autosomal recessive lysosomal storage disease characterized by deficient activity of the enzyme alpha-D-mannosidase. There is a wide range of signs and symptoms including hepatomegaly, splenomegaly, hearing loss, respiratory infections, mental retardation, skeletal abnormalities, leveled nasal bridge and protruding forehead.

Qeios ID: 6HBKCT · https://doi.org/10.32388/6HBKCT