

Open Peer Review on Qeios

Mucolipidosis Type IIIA

National Cancer Institute

Source

National Cancer Institute. <u>Mucolipidosis Type IIIA</u>. NCI Thesaurus. Code C125595.

A lysosomal storage disease characterized by multiple bone formation abnormalities, progressive joint stiffness, developmental abnormalities, hearing loss, hepatosplenomegaly, increased acne, enlarged tongue, and cornea clouding due to accumulation of lipid substances.

Qeios ID: WZ2J4T · https://doi.org/10.32388/WZ2J4T