Deafness, Autosomal Recessive 1A

National Cancer Institute

Source

National Cancer Institute. Deafness, Autosomal Recessive 1A. NCI Thesaurus. Code C129022.

An autosomal recessive disorder caused by mutations in the GJB2 gene, encoding gap junction beta-2 protein. The condition is characterized by profound sensorineural hearing loss and may be associated with vestibular dysfunction.