

Open Peer Review on Qeios

Deafness, Autosomal Recessive 4, with Enlarged Vestibular Aqueduct

National Cancer Institute

Source

National Cancer Institute. <u>Deafness, Autosomal Recessive 4, with Enlarged Vestibular</u>
<u>Aqueduct</u>. NCI Thesaurus. Code C158787.

An autosomal recessive condition caused by mutation(s) in one of several genes, most often SLC26A4 encoding pendrin. It is characterized by hearing loss and enlargement of the vestibular aqueduct. Mutation(s) in the SLC26A4 gene also cause Pendred syndrome.

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