Deafness, Autosomal Recessive 4, with Enlarged Vestibular Aqueduct

Source


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.