

Pendrin is an anion transporter that is expressed in several organs. In the thyroid gland, pendrin is localized at the apical pole of thyrocytes and it is responsible for the iodide efflux from thyrocytes into the colloid in the follicular lumen where iodide is organificated. The extrathyroidal expression was shown in the inner ear, kidney, placenta and mammary gland. Carriers of mutations in the pendrin gene (PDS, SLC26A4) display variable phenotypical features following the autosomal recessive manner of the inheritance: combined thyroid and hearing affection (Pendred syndrome - OMIM274600), nonsyndromic autosomal recessive neurosensory deafness (DFNB4 - OMIM600791) or isolated enlarged vestibular aqueduct (EVA - OMIM603545). The thyroid affection is usually manifested as euthyroid or hypothyroid goitre in the second decade of life. In a minority of patients, dyshormonogenesis is present at birth, and the disease is diagnosed in the frame of the nation-wide neonatal screening for congenital hypothyroidism.