Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome. ORPHA:71267

Dentinogenesis imperfecta-short stature-hearing loss-intellectual disability syndrome is characterised by the association of dentinogenesis imperfecta, delayed tooth eruption, facial dysmorphology, small stature, sensorineural hearing loss and mild intellectual deficit. It has been described in two brothers born to consanguineous parents. Transmission is autosomal recessive.