Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome

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Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome. ORPHA:397618

Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome is a rare, genetic, eye disease characterized by foveal hypoplasia, optic nerve misrouting with an increased number of axons decussating at the optic chiasm and innervating the contralateral cortex, and posterior embryotoxon or Axenfeld anomaly (indicating anterior segment dysgenesis), in the absence of albinism. Patients present congenital nystagmus, decreased visual acuity, refractive errors and, ocassionally, strabismus. Microphthalmia and retinochoroidal coloboma may also be associated.