Hartsfield syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hartsfield syndrome. ORPHA:2117

Hartsfield syndrome is a rare, genetic, developmental defect during embryogenesis malformation syndrome characterized by the association of variable degrees of holoprosencephaly and uni- or bilateral ectrodactyly of the hands and/or feet. Additional variable features, including facial dysmorphism (e.g. hypertelorism, short bulbous nose, long philtrum, dysplastic/low-set ears, cleft lip and palate, tented upper lip), other brain malformations (such as corpus callosum agenesis, absent septum pellucidum, absent olfactory bulbs/tracts, vermian hypoplasia), pituitary gland-related endocrine disorders (e.g. central diabetes insipidus, hypogonadotropic hypogonadism) and hypothalamic dysfunction, may be associated.