Brachytelephalangy-dysmorphism-Kallmann syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Brachytelephalangy-dysmorphism-Kallmann syndrome. ORPHA:1295

Brachytelephalangy - dysmorphism - Kallmann syndrome is a developmental anomaly characterized by brachytelephalangy, distinct craniofacial features (prominent square forehead, telecanthus, small nose, malar hypoplasia, smooth philtrum and thin upper lip), and relative to other family members, a short stature. These features may be associated with anosmia and hypogonadotropic hypogonadism (considered as Kallman syndrome ; see this term). Brachytelephalangy - dysmorphism - Kallmann syndrome has been described in a mother and her son and there have been no further descriptions in the literature since 1986.