Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome. ORPHA:404476

Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome is a rare, genetic, overgrowth syndrome characterized by global developmental delay, macrosomia with subsequent somatic overgrowth, bilateral cystic lung lesions, congenital nephromegaly and bilateral Wilms tumor. Craniofacial dysmorphism includes macrocephaly, frontal bossing, large anterior fontanelle, mild hypertelorism, ear pit, flat nasal bridge, anteverted nares and mild micrognathia. Additional features may include brain and skeletal anomalies, enlarged protuberant abdomen, fat pads on dorsum of feet and toes, and rugated soles with skin folds, as well as umbilical/inguinal hernia and autistic behavior.