BRESEK syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. BRESEK syndrome. ORPHA:85284

X-linked mental retardation, Reish type is characterised by Brain anomalies, severe mental Retardation, Ectodermal dysplasia, Skeletal deformities (vertebral anomalies, scoliosis, polydactyly), Ear/eye anomalies (maldevelopment, small optic nerves, low set and large ears with hearing loss) and Kidney dysplasia/hypoplasia (giving the acronym BRESEK syndrome).