

Opis choroby *

Definicja

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).

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	BRESHECK syndrome
	Zespół BRESHECK

Kod ORPHA	Kod OMIM	Kod ICD10
85284	308205	Q87.8

Kod ICD11
LD90

*Źródło

orphanet