DISEASE:
BRESEK syndrome

NAME:	BRESEK syndrome
DESCRIPTION:	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).
ORPHACODE:	85284
SYNONYMS:	BRESHECK syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MBTPS2
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RELATED CONTENT

Related Genetic Tests

- Ichthyosis (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- membrane bound transcription factor peptidase, site 2

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Ichthyosis and erythroderma (98 genes) - KUL