DISEASE:
Leigh syndrome with nephrotic syndrome

NAME:	Leigh syndrome with nephrotic syndrome
DESCRIPTION:	A rare, genetic neurometabolic disease characterized by encephalomyopathy (including developmental delay, nystagmus, progressive ataxia, dystonia, amyotrophy, visual loss, sensorineural deafness, seizures) and bilateral, symmetrical lesions in the basal ganglia or brainstem on imaging, associated with nephrotic syndrome.
ORPHACODE:	255249
SYNONYMS:	Infantile subacute necrotizing encephalopathy with nephrotic syndrome Leigh disease with nephrotic syndrome
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	COQ2 PDSS2
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RELATED CONTENT

Related Genetic Tests

- Leigh / NARP Syndrome
- Mitochondrial disorders (gene panel)
- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- coenzyme Q2, polyprenyltransferase
- decaprenyl diphosphate synthase subunit 2

Related Gene Panels

- Leigh syndrome (mtDNA / 37 genes) - VUB
- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG
- mitochondrial disease, nuclear based (343 genes) - VUB

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