DISEASE:
LAMB2-related infantile-onset nephrotic syndrome

NAME:	LAMB2-related infantile-onset nephrotic syndrome
DESCRIPTION:	LAMB2-related infantile-onset nephrotic syndrome is a rare primary glomerular disease due to homozygous mutations in LAMB2 gene, characterized by prenatal or early-onset progressive steroid-resistant nephrotic syndrome leading to renal failure, and variable ocular defects including myopia, fundus abnormalities, strabismus or nystagmus, without severe visual impairment or blindness. Patients present in early infancy with massive proteinuria, edema, hypertension, and hyperlipidemia. Psychomotor development is normal.
ORPHACODE:	306507
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	LAMB2
CREATED:	13 May 2019 - 01:02
CHANGED:	01 Jul 2019 - 06:57

RELATED CONTENT

Related Genetic Tests

- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- laminin subunit beta 2

Related Gene Panels

- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG