

**DISEASE:**  
**Galloway-Mowat syndrome**

<b>NAME:</b>	Galloway-Mowat syndrome
<b>DESCRIPTION:</b>	A rare, genetic multisystem disorder characterized by a neurodegenerative disorder associating global developmental delay, progressive microcephaly, and progressive cerebral and cerebellar atrophy with extrapyramidal involvement, progressive optic atrophy, and in many patients early-onset steroid-resistant nephrotic syndrome.
<b>ORPHACODE:</b>	2065
<b>SYNONYMS:</b>	Galloway syndrome Microcephaly-hiatus hernia-nephrotic syndrome Nephrosis-neuronal dysmigration syndrome

<b>XREF(S):</b>	<u>Orphanet</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u> <u>MeSH</u> <u>ICD-10</u> <u>OMIM</u> <u>OMIM</u> <u>OMIM</u>
<b>ANALYTE(S):</b>	<u>OSGEP</u> <u>TP53RK</u> <u>WDR73</u> <u>NUP107</u> <u>TPRKB</u> <u>LAGE3</u> <u>WDR4</u> <u>GON7</u> <u>NUP133</u> <u>YRDC</u>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### 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

- GON7 subunit of KEOPS complex
- L antigen family member 3
- nucleoporin 107
- nucleoporin 133
- O-sialoglycoprotein endopeptidase
- TP53 regulating kinase
- TP53RK binding protein
- WD repeat domain 4
- WD repeat domain 73
- yrdC N6-threonylcarbamoyltransferase domain containing

### Related Gene Panels

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

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