

Full name:	Metabolic diseases with hepatic disorders (20 genes) - UCL
Abbreviation:	Panel GP
Type of panel:	<u>Custom panel</u>
Laboratory:	<u>Centre de Génétique Médicale UCL</u>
Created:	06 Nov 2019 - 14:45
Changed:	02 Feb 2022 - 11:28

Related Diseases

- Carnitine palmitoyl transferase 1A deficiency
- Cerebrotendinous xanthomatosis
- Glycogen storage disease due to glycogen branching enzyme deficiency, progressive hepatic form
- Mucopolysaccharidosis type 7
- NON RARE IN EUROPE: Heterozygous familial hypercholesterolemia
- Neonatal intrahepatic cholestasis due to citrin deficiency
- Smith-Lemli-Opitz syndrome
- Transaldolase deficiency
- Wilson disease

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ATP7B</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>CPT1A</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>CYP27A1</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>DGUOK</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>DHCR7</u>	100.00	1	Part of Custom Gastro-pneumo panel

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>EHHADH</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>GBE1</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>GNAS</u>	0.00	1	Part of Custom Gastro-pneumo panel / only Exons 8 and 9
<u>GUSB</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>LIPA</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>MPV17</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>NEU1</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>NPC1</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>NPC2</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>POLG</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>SI</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>SLC25A13</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>SMPD1</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>TALDO1</u>	100.00	1	Part of Custom Gastro-pneumo panel
<u>TRMU</u>	100.00	1	Part of Custom Gastro-pneumo panel