

GENETIC TEST:
Medium chain acyl-CoA dehydrogenase deficiency (MCAD hot spot mutation - p.Lys329Glu)

FULL NAME:	Medium chain acyl-CoA dehydrogenase deficiency (MCAD hot spot mutation - p.Lys329Glu)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Post-natal Diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Amniotic fluid, Chorionic villi
METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	PCR based technique
RIZIV CODE:	565390-565401
TURNAROUND TIME (MAXIMUM):	10 days
CREATED:	02 Sep 2019 - 07:55

CHANGED:	11 Dec 2023 - 15:13
URL:	https://www.chuliege.be/jcms/c_525401/fr/deficit-en-medium-chain-coa-dehydrogen...

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