

Molecular genetic analyses requisition

Patient identification	
Date of birth	
Name	
Gender: male	female

Patient informed consent for genetic testing is required.

Requesting information's
Requesting physician
Billing to (If different from requesting physician)
VAT nr.
Answer to (If different from requesting physician)

Test Sample	
Date	Time

Family with known mutations	
Gene	Mutation
Index patient	
Name:	
Date of birth:	
Other information	

Tests			
Disease	Gene	Screen	Known
Medium-chain acyl-CoA dehydrogenase (MCAD) defect [#]	ACADM		
Very-long-chain acyl-CoA dehydrogenase (VLCAD) defect [#]	ACADVL		
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) defect [#]	HADHA		
Mitochondrial Trifunctional Protein (MTP) defect [#]	HADHA HADHB		
Isovaleryl-CoA dehydrogenase (IVD) defect or Isovaleric acidemia (IVA) [#]	IVD		
Short-chain acyl-CoA dehydrogenase (SCAD) defect	ACADS		
Multipel acyl-CoA dehydrogenation defect (MADD)	ETFA ETFB ETFDH SLC52A1 SLC52A2 SLC52A3 RFK FLAD1 MFT		
Carnitine palmitoyltransferase 1A (CPT1A) defect	CPT1A		
Carnitine palmitoyltransferase 2 (CPT2) defect	CPT2		
Acyl-CoA dehydrogenase 9 (ACAD9) defect	ACAD9		
Isobutyryl-CoA dehydrogenase (IBD) defect	ACAD8		
Short-branched-chain acyl-CoA dehydrogenase (SBCAD) defect	ACADSB		
Ethylmalonyl-CoA decarboxylase 1 (ECHDC1) defect	ECHDC1		
Ethylmalonic encephalopathy 1 (ETHE1) defect	ETHE1		
Thiosulfate sulfurtransferase (TST) defect	TST		
Sulfide quinone oxidoreductase (SQOR) defect	SQOR		
Heat shock protein 10 (Hsp10) defect	HSPE1		
Heat shock protein 60 (Hsp60) defect	HSPD1		

[#] Accredited analysis

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How to handle the test sample

Before sending, any samples please contact MMF. Telephone number: +45 7845 5412
Other contact information can be found at: www.clin.au.dk/mmf

Sample material. Blood spot, DNA or EDTA blood can be used. Samples should be marked with name and birth date

Durability: There are no durability issues with Blood spot and DNA samples. The EDTA blood sample is stable at room temperature for 4 days. Storing the sample for more than 4 days at room temperature will decrease the quality of the sample, but we will still try to analyze it.

Informed consent. In order to do molecular genetic analysis it is assumed that the patient is informed and has given permission for the analysis

How to send the samples. Ordinary mail to Molekylær Medicinsk Forskningsenhed (MMF), Aarhus Universitetshospital, Palle Juul-Jensens Boulevard, 8200 Aarhus N, Denmark

Clinical and biochemical informations