

Disorder	Site	Metabolite/ Gene	Test	Sample Type	TAT	Contact	Email address	Tel Number	Courier Requirements	
Glutaric aciduria type 1 (GA1)	Birmingham	GCDH	DNA sequencing test not available	-	-	-	-	-	-	
		Urine Quant 3-OH glutarate	-	Dried bloodspot	-	-	-	-	-	
		Glut CoA DH	-	-	-	-	-	-	-	
	Great Ormond Street	GCDH	DNA sequencing test not available	N/A	N/A	N/A	-	-	-	N/A
		Urine Quant 3-OH glutarate	Urine Organic acid (semiquantitative)	Urine (freeze immediately)	2-4 weeks	Helen Prunty	helen.prunty@gosh.nhs.uk	0207 7813 8319	Send frozen	
		Bloodspot glutarylcarnitine	Full scan acylcarnitine profile	Dried blood spot (4 spots)	1-2 weeks	-	-	-	Send by 1st class post	
		Glut CoA DH	Enzyme assay not available	N/A	N/A	-	-	-	N/A	
	Guy's & St Thomas'	GCDH	Mutation screen of coding exons and intron-exon boundaries (exons 2-12)	Dried bloodspot or extracted DNA	2 Weeks	David Cregeen	david.cregeen@viapath.co.uk	020 7188 2591	-	
		Urine Quant 3-OH glutarate	N/A	N/A	N/A	-	-	-	N/A	
		Glut CoA DH	N/A	N/A	N/A	-	-	-	N/A	
	Leeds	GCDH	DNA sequencing test not available	N/A	N/A	N/A	N/A	N/A	N/A	
		Urine Quant 3-OH glutarate	N/A	N/A	N/A	N/A	N/A	N/A	N/A	
		Glut CoA DH	N/A	N/A	N/A	N/A	N/A	N/A	N/A	
	Manchester	GCDH	DNA sequencing test not available	N/A	N/A	N/A	N/A	N/A	N/A	
Urine Quant 3-OH glutarate		Test are not available	N/A	N/A	N/A	N/A	N/A	N/A		
Glut CoA DH		Test are not available	N/A	N/A	N/A	N/A	N/A	N/A		
Sheffield	GCDH	Mutation screen of coding exons and intron-exon boundaries (exons 2-12)	Dried bloodspot or extracted DNA	2 Weeks	Richard Kirk	richard.kirk@sch.nhs.uk	0114 271 7003	Send by 1st class post		
	Urine Quant 3-OH glutarate	Test not available	N/A	N/A	N/A	N/A	N/A	N/A		
	Glut CoA DH	Enzyme assay	fibroblasts	6 weeks	Simon Olpin	simon.olpin@sch.nhs.uk	0114 271 7267	Send by 1st class post		
Isovaleric acidaemia (IVA)	Birmingham	-	-	-	-	-	-	-	-	
	Great Ormond Street	N/A	DNA sequencing test is not available	N/A	N/A	-	-	-	N/A	
		Urine Quant isovalerylglycine	Urine organic acid (semiquantitative)	Urine (freeze immediately)	2-4 weeks	Helen Prunty	helen.prunty@gosh.nhs.uk	0207 7813 8319	Send frozen	
		Bloodspot isovalerylcarnitine	Full scan acylcarnitine profile	Dried blood spot (4 spots)	1-2 weeks	-	-	-	Send by 1st class post	
	Guy's & St Thomas'	IVD	Benign variant c.941C.T (p.Ala314Val) also known as c.932.T (p.A282V) sequence analysis	Dried bloodspot or extracted DNA	1 Week	David Cregeen	david.cregeen@viapath.co.uk	020 7188 2591	-	
			Mutation screen of coding exons and intron-exon boundaries (exons 2-12)		2 Weeks					
	Leeds	N/A	DNA sequencing test not available	N/A	N/A	N/A	N/A	N/A	N/A	
Sheffield	IVD	Benign variant c.941C.T (p.Ala314Val) also known as c.932.T (p.A282V) sequence analysis	Dried bloodspot or extracted DNA	3 Working Days	Richard Kirk	richard.kirk@sch.nhs.uk	0114 271 7003	Send by 1st class post		
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)	Birmingham	-	-	-	-	-	-	-	-	
	Great Ormond Street	HADHA	Common variant c.1528G>C (p.Glu510Gln) PCR and restriction enzyme analysis	Dried bloodspot or extracted DNA	3 days	Sam Loughlin	sam.loughlin@gosh.nhs.uk	020 7762 6888	-	
			Hydroxyacylcarnitine compounds	Full scan acylcarnitine profile	Dried blood spot (4 spots)	1-2 weeks	Helen Prunty	helen.prunty@gosh.nhs.uk	0207 7813 8319	Send by 1st class post
	Guy's & St Thomas'	HADHA	Common variant c.1528G>C (p.Glu510Gln) (nested) PCR and restriction enzyme analysis	Dried bloodspot or extracted DNA	3 Days	David Cregeen	david.cregeen@viapath.co.uk	020 7188 2591	-	
	Leeds	N/A	DNA sequencing test not available	N/A	N/A	N/A	N/A	N/A	N/A	
	Sheffield	HADHA	Common variant c.1528G>C (p.Glu510Gln) (nested) PCR and restriction enzyme analysis	Dried bloodspot or extracted DNA	3 Working Days	Richard Kirk	richard.kirk@sch.nhs.uk	0114 271 7003	Send by 1st class post	
Maple Syrup Urine Disease (MSUD)	Birmingham	Alloisoleucine	-	Dried bloodspot	24 hrs	-	-	-	-	
		Leucine oxidation	-	-	-	-	-	-	-	
	Great Ormond Street	Alloisoleucine	Branched chain amino acids	Dried bloodspot	4 days	Helen Prunty	helen.prunty@gosh.nhs.uk	0207 7813 8319	Send by 1st class post	
		Leucine oxidation	-	-	-	-	-	-	-	
	Guy's & St Thomas'	Alloisoleucine	Available	Dried bloodspot	24 hrs	David Cregeen	david.cregeen@viapath.co.uk	020 7188 2591	-	
		Various	Cultured cell assay of leucine decarboxylating system	Skin biopsy or cultured fibroblasts	6 Weeks*	-	-	-	-	
	Leeds	Alloisoleucine	Available	Dried bloodspot	24 hrs	Daniel Herrera	daniel.herrera@leedsth.nhs.uk	0113 20 64256	Send by 1st class post	
		Leucine oxidation	Test Not Available	N/A	N/A	N/A	N/A	N/A	N/A	
	Manchester	Alloisoleucine	Available	Dried bloodspot	24 hrs	-	-	-	-	
		Leucine oxidation	Test not available	N/A	N/A	N/A	N/A	N/A	N/A	
Sheffield	Alloisoleucine	Available	Dried bloodspot	24 hrs	Jennifer Watkinson	Jennifer.Watkinson@sch.nhs.uk	0114 271 7445	Send by 1st class post		
	Leucine oxidation	Cultured cell assay of leucine decarboxylating system	Cultured fibroblasts	6 weeks	Simon Olpin	simon.olpin@sch.nhs.uk	0114 271 7267	Send by 1st class post		
Homocystinuria (HCU)	Birmingham	Methionine	-	Dried bloodspot	-	-	-	-	-	
	Great Ormond Street	Methionine	Plasma amino acids	0.2 ml li hep plasma (Separate ASAP. Freeze immediately.)	1-2 weeks	Helen Prunty	helen.prunty@gosh.nhs.uk	0207 7813 8319	Send frozen	
		Total Homocysteine	Plasma amino acids	0.2 ml li hep plasma (Separate ASAP. Freeze immediately.)	-	-	-	-	-	
	Guy's & St Thomas'	Methionine	Available	Dried bloodspot	-	-	-	020 7188 2591	-	
	Leeds	N/A	Test not available	N/A	N/A	N/A	N/A	N/A	N/A	
	Sheffield	N/A	Test not available	N/A	N/A	N/A	N/A	N/A	N/A	

* from cultured cells. Culture from skin biopsy may take an additional 5 to 6 weeks

Dutch Diagnostic Services	
Diagnostic testing	Enzyme activity measurement of LCHAD/MTP in isolated lymphocytes & cultured skin fibroblasts
Shipping Arrangements	Send 5-7 ml of EDTA blood at room temperature, please include a control blood sample The shipment should arrive within 24-48 hrs after drawing Alternatively, isolated lymphocytes of the patient and 1 control subject can be shipped on dry ice Samples should arrive MONDAY to THURSDAY (The lab is open from 8.30 to 17.00) (The lab is open on Friday, but in their experience regularly shipments by couriers are delayed and arrive after opening hours or even a day later)
Shipping Documentation	Visit the website www.labgmd.nl -<http://www.labgmd.nl> for the forms to be sent with the shipment (http://www.labgmd.nl/pdf/gmd_enzen.pdf) and the shipment instructions (There is an address label on this form that can be used)
Testing Costs	350 Euro for LCHAD activity measurement 350 Euro for long-chain 3-ketoacyl-CoA thiolase activity measurement 50 Euro for lymphocyte isolation 100 Euro fibroblast culture Total 750 Euro for the analysis in lymphocytes and 800 Euro in fibroblasts
Contact details	Email address: gmz_enzym@amc.nl Phone Number: +31 20 5662026/ 5393