

## TURNAROUND TIMES OF DIAGNOSTIC ANALYSES AT THE TRANSLATIONAL METABOLIC LABORATORY (TML)

For more information, please contact the laboratory: tel. +31 (0)24-36 14567 (administrative office).

Category	Analyses group	Turnaround time	Frequency	Remark
<b>Metabolite diagnostics</b>	Metabolite investigation in case of suspicion metabolic disease	4 - 6 weeks	Depending on analyses: weekly to 1x per 3 weeks	Depending on the question and material type. In consultation with the laboratory specialist an investigation can be performed with priority, if the clinical condition of the patient requires this. Interim results can also be requested from a laboratory specialist.
	Phenylalanine control therapy	1 week	Weekly on wednesday	Arrival before Wednesday, result in the same week.
	Methylmalonic acid (MMA)	3 weeks	1x per 2-3 weeks	
	Homocysteine	2-3 weeks	1x per 1-2 weeks	
<b>Complement Mediated diseases</b>	C3bBbP, C3bc and C5b-9	6 weeks	Every 4 weeks	Special appointment possible.
	Eculizumab (EM)	2 to 4 weeks	weekly	Special appointments possible.
	EM inhibiting capacity and EM-C5	6 weeks	Every 4 weeks	These actions are only used with an eculizumab free levels between 8 - 100 µg / ml.

	Anti-FH, anti-LPS	6 weeks	Every 4 weeks	Special appointment possible.
	C3Nef, C4Nef, C4D	6 weeks	Every 4 weeks	Special appointment possible.
	Factor H , Factor I	6 weeks	Every 4 weeks	Special appointment possible.
<b>Glycosylation diseases</b>	Mass Spectrometry Transferrin (instead of TIEF)	4-6 weeks	Every 2 weeks	
	apoCIII	6 weeks	Every 3 weeks	
	Other	6-8 weeks	On offer	Depending on the question and material type.
	Glycomics in plasma			In consultation.
<b>Lysosomal enzyme diagnostics</b>	Regular enzyme diagnostics (Glycosphingolipidose and oligosaccharidose enzymes)	5 weeks	Every 3 weeks	This applies to blood samples (heparin whole blood) and is exclusive possible growing time for fibroblasts. Depending on question and material type.
	Complete mucopolysaccharidosis package	100 days	On offer	
	Specific enzyme requests (eg based on strong clinical / biochemical evidence and with regard to therapeutic possibilities)			In consultation.
	Pseudocholesterase measurement (including dibucaine number)	100 days	On offer	Depending on question and material type.
<b>Mitochondrial enzyme diagnostics</b>	All requests	2 months		Depending on the question and material type. With materials. Fibroblasts and skin biopsy the cell growth rate should be taken into account (average duration 4-6 weeks).
	Q10	3 months	On offer	

	Ca-ATPase	3 months	On offer	
<b>DNA diagnostics</b>	All gene/gene package requests, MLPA requests	8 weeks		Depending on the question and material type. In consultation with the laboratory specialist investigation can be performed with priority if the clinical condition of the patient requires this.
	Segregation analysis (mutation analysis)	4 weeks		
	Mitochondrial DNA testing	8 weeks		
	Prenatal testing	2 weeks		
	Depletion and deletion test	100 days	On offer	
	NGS (WES diagnostics)	100 days		Only concerns gene package research (mitochondrial, metabolic, iron metabolism).
	WES (outside the above mentioned gene packages)			Only with involvement of clinical geneticist.
<b>Functional Genomics</b>	Functional Genomics	6 months	On offer	Always in consultation (no patient diagnostics).
<b>Iron metabolism</b>	Hepcidin determination	3 weeks	Every 3 weeks	
<b>Neurochemistry diagnostics</b>	Pterine in urine	4 weeks	Every 3 weeks	
	Pterine in liquor	4 weeks	Every 3 weeks	
	Neurotransmitter metabolite in liquor (HVA, 5-HIAA, MHPG, 3-methoxy-Tyrosine, L-dopa)	4 weeks	Every 2 or 3 weeks	
	5 MTHF in liquor	4 weeks	Every 3 weeks	

	HVA in blood	4 weeks	Every week	Emergency diagnostics only in consultation.
	HVA in urine	2 weeks	Every week	Emergency diagnostics only in consultation.
	5-HIAA in blood	4 weeks	Every week	
	5-HIAA in urine	2 weeks	Every week	
	VMA in urine	2 weeks	Every week	Emergency diagnostics only in consultation.
	Aromatic L-aminodecarboxylase assay in plasma / serum		In consultation	Only in consultation (+/- 5 weeks).
	Dopamine-beta-hydroxylase (DBH) in serum/	Upon receipt	In consultation	
	Serotonin (5-HT) in serum	4 weeks	Every 3 weeks	
	Free kappa chain oligoclonal IGG bands in liquor and serum	3 weeks	Every 2 weeks	
	Anti-ganglioside (GM1, GM2, GQ1b) antibodies in serum	4 weeks	Every 4 weeks	
	Anti-ganglioside (GM1, GM2, GQ1b) antibodies in liquor	4 weeks		
	$\beta$ 2-transferrin in body material (nose / ear fluid)	4 days	2 x per week	Cyto: 1 day upon receipt before noon.
	Paraneoplastic antibodies in serum and liquor	10 days	Within 3 days after receipt	
	Oligoclonal IgG bands in liquor and serum	10 days	2 x per week	
	14-3-3 protein in liquor	3 weeks	Every 2 weeks	With holidays up to 3 weeks.
	GFAP in liquor and blood	4 weeks	Every 2 weeks	With holidays up to 3 weeks.

	MPB in liquor	4 weeks	Every 2 weeks	With holidays up to 3 weeks.
	CXCL 13 in Liquor	3 weeks	Every 2 weeks	
	NFL in liquor	4 weeks	Elke 4 weken	
	Amyloid beta 42 in liquor	3 weeks	Every 2 weeks	With holidays up to 3 weeks.
	Phospho tau in liquor	3 weeks	Every 2 weeks	With holidays up to 3 weeks.
	h-tau in liquor	3 weeks	Every 2 weeks	With holidays up to 3 weeks.
	Amyloid beta 40 in liquor	4 weeks	Every 2 weeks	With holidays up to 3 weeks.

