

INHERITED METABOLIC DISORDERS SPECIAL INVESTIGATIONS REQUIRED URGENTLY

Aim: To provide guidance for laboratories across Scotland regarding expected turnaround times for urgent investigations in suspected metabolic disorders

Analyte	Urgency	Turnaround time (All urgent requests MUST BE DISCUSSED with laboratory staff)
Ammonia	+++	2 hours
Lactate*	+++	2 hours
Creatine kinase	+++	4 hours
Urine Organic acids	++	1-2 working days
Plasma amino acids	++	2 working days
Phenylalanine [#]	++	2 working days
Gal-1-PUT	++	2 working days
NEFA (FFA)	+	4 working days
Acylcarnitines	+	4 working days
Beta-hydroxybutyrate	+	4 working days
Urine orotic acid	+	4 working days
Insulin ^{\$}	+	4 working days

OR: +++ = “now, please...”
 ++ = “late tomorrow or the day after would be great, thanks...”
 + = “end of the week or early next week is fine...”

* *Lactate* can be measured by near-patient testing on blood gas analysers.

- # *Phenylalanine*: clinical referral guidelines and standards for phenylketonuria state: “Results of confirmatory diagnostic tests should be available within 24 hours of being taken and ideally on the same visit.”
- \$ *Insulin*: interpretation of insulin result requires paired glucose sample.



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This guidance is not intended to be construed or to serve as a standard of care. Standards of care are determined on the basis of all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from national guidance or any local guidelines derived from it should be fully documented in the patient's case notes at the time the relevant decision is taken.