

Specialist Portfolio Clinical Chemistry Competence – 7.16 Gastrointestinal Inherited Metabolic Disorders and Newborn Screening: Prenatal Screening for Down’s Syndrome

Name:

To be signed off by appropriately qualified staff after satisfactory performance observed and knowledge displayed

	Trainer		
	Name	Grade	Signature
Explain the clinical manifestation of at least one inherited metabolic disease resulting from: <ul style="list-style-type: none"> • Decreased synthesis of a normal metabolite; • Increased synthesis of a normal metabolite; • Compromised trans-membrane transport; • Decreased receptor synthesis; • Altered binding of a coenzyme to an enzyme 			
Confirm times when the samples should be collected			

Explain the methods available and the associated advantages and disadvantages for markers of the most common inborn metabolic disorders			
Analyse results, consider possible interference, and take appropriate action			
Report results appropriately to the significance of the test			
Monitor results, consider possible interference, and take appropriate action			
Report results appropriately to the significance of the result			
Complete all relevant documentation in accordance with quality control and audit requirements			