



METABOLIC AND NEWBORN SCREENING LABORATORY

Lab / Clinic	Test	Test Synonyms	Specimen Requirements	Cost \$AU	Typical turn around time (receipt of sample to results available)	Notes
Metabolic screening	3-hydroxyglutaric		At least 5 mL of urine, store and transport frozen	\$100	Up to 21 days	Diagnosis of glutaric aciduria type 1
Metabolic screening	7-dehydrocholesterol	Sterols	1 mL of blood in heparin (orange top) tube. Plasma must be separated within 30 minutes of collection and plasma stored frozen and wrapped in foil.	\$150	Up to 21 days	Diagnosis of Smith-Lemli-Opitz syndrome
Metabolic screening	Acetoacetate		1 ml of blood in special tube - contact lab	\$20	Up to 21 days	
Metabolic screening	Acyl carnitine		Newborn screening (Guthrie) card. After collection, wrap the card in paper towelling and staple to the request card. Do not place in plastic sample bags until the card is completely dry (at least 4 hours).	\$60	1 to 3 days. Urgent testing can be performed but must be arranged with the metabolic physician.	
Metabolic screening	Adelaide enzymes	Lysosomal enzymes	10 mL of blood in EDTA (red top) tube. 5 mL of blood is generally suitable for younger children. Must be collected on Tuesday or Wednesday before 10AM. Do not separate blood, store at 4C.			Analysed at Women's and Children's Hospital, Adelaide. External organisations should send their samples directly - we do not accept samples from external organisations.
Metabolic screening	Amino acids - blood		Patient should preferably be fasted. At least 1 mL of blood in a heparin tube (orange top tube) cooled on wet ice. Separate plasma within 1 hour of collection and store and transport frozen.		2 to 6 days. Urgent testing can be performed but must be arranged with the metabolic physician.	
Metabolic screening	Amino acids - CSF		0.5 mL of clear CSF in plain tube, snap frozen on dry ice.		2 to 6 days. Urgent testing can be performed but must be arranged with the metabolic physician.	
Metabolic screening	Amino acids - urine		At least 5 mL of urine, store and transport frozen	\$100	1 to 4 days. Urgent testing can be performed the same day but must be arranged with the metabolic physician.	Part of metabolic screen
Metabolic screening	Bile acid profile		At least 5 mL of urine, store and transport frozen	\$350	Up to 21 days	
Metabolic screening	Biotinidase screen		Newborn screening (Guthrie) card. After collection, wrap the card in paper towelling and staple to the request card. Do not place in plastic sample bags until the card is completely dry (at least 4 hours).			Analysed at NSW Biochemical Genetics. External organisations should send their samples directly - we do not accept samples from external organisations.

Current as at February 2007

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Metabolic screening	Carnitine, free and total		1 mL of blood in heparin (orange top) tube. Separate plasma and store and transport frozen.	\$83	Up to 21 days	
Metabolic screening	Chitotriosidase		1 mL of blood in serum gel (brown top) tube. Must be collected on Tuesday or Wednesday before 10AM. Do not separate blood, store at 4C.			Analysed at Women's and Children's Hospital, Adelaide. External organisations should send their samples directly - we do not accept samples from external organisations.
Metabolic screening	Cystine		At least 5 mL of urine, store and transport frozen	\$100	1 to 4 days. Urgent testing can be performed the same day but must be arranged with the metabolic physician.	
Metabolic screening	Free fatty acids		At least 1 mL of blood in a fluoride/oxalate (grey top) tube. Separate plasma and store and transport frozen.	\$35	Up to 21 days	
Metabolic screening	Galactoscreen		At least 0.5 mL blood in a heparin (orange top) tube. Collect whole blood specimen prior to any blood transfusion. Do not separate blood. Store and transport at 4°C.	\$52	Same day	
Metabolic screening	Hydroxy butyrate		At least 1 mL of blood in a fluoride/oxalate (grey top) tube. Separate plasma and store and transport frozen.	\$20	Up to 21 days	
Metabolic screening	Metabolic screen		At least 5 mL of urine, store and transport frozen	\$100	1 to 4 days. Urgent testing can be performed the same day but must be arranged with the metabolic physician.	Standard urine metabolic screen includes total glycosaminoglycans, amino acids and acyl carnitines. Organic acids and other tests may be included depending on clinical information provided or a specific request.
Metabolic screening	Mucopolysaccharide screen		At least 10 mL of urine, early morning preferred, store and transport frozen	\$100	Up to 21 days	
Metabolic screening	Oligosaccharides		At least 10 mL of urine, early morning preferred, store and transport frozen			Analysed at Women's and Children's Hospital, Adelaide. External organisations should send their samples directly - we do not accept samples from external
Metabolic screening	Orotic acid		At least 5 mL of urine, store and transport frozen	\$75	Up to 21 days	

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Metabolic screening	Phytanic acid		3 ml of blood in EDTA (red top) or heparin (orange top) tube.			Analysed at Women's and Children's Hospital, Adelaide. External organisations should send their samples directly - we do not accept samples from external organisations.
Metabolic screening	Pompe disease		Newborn screening (Guthrie) card. After collection, wrap the card in paper towelling and staple to the request card. Do not place in plastic sample bags until the card is completely dry (at least 4 hours).			Analysed at Women's and Children's Hospital, Adelaide. External organisations should send their samples directly - we do not accept samples from external organisations.
Metabolic screening	Purine and pyrimidine screen		At least 5 mL of urine, store and transport frozen	\$54	Up to 21 days	
Metabolic screening	Pyruvate - blood		1 ml of blood in special tube - contact lab	\$20	Up to 21 days	Generally paired with lactate
Metabolic screening	Pyruvate - CSF		0.5 mL of clear CSF in plain tube, snap frozen on dry ice.	\$20	Up to 21 days	Generally paired with lactate
Metabolic screening	Reducing substances		At least 5 mL of urine, store and transport frozen		1 to 4 days. Urgent testing can be performed the same day but must be arranged with the metabolic physician.	
Metabolic screening	Succinyl acetone		At least 5 mL of urine, store and transport frozen	\$100	Up to 21 days	Diagnosis of tyrosinaemia type 1
Metabolic screening	Transferrin isoforms		At least 2 mL of blood in a serum gel (brown top) tube. Separate and freeze serum for storage and transport.	\$140	Up to 21 days	Diagnosis of congenital disorders of glycosylation
Metabolic screening	Very long chain fatty acids	VLCFA	At least 2 mLs of blood in a heparin (orange top) or EDTA tube. Separate and freeze plasma for storage and transport.			Analysed at Women's and Children's Hospital, Adelaide. External organisations should send their samples directly - we do not accept samples from external organisations.
Metabolic screening	Organic acids		At least 5 mL of urine, store and transport frozen	\$100	1 to 4 days. Urgent testing can be performed the same day but must be arranged with the metabolic physician.	Part of metabolic screen

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Metabolic screening	Pipecolic		At least 5 mL of urine, store and transport frozen	\$100	Up to 21 days	
Metabolic screening	Methylmalonic acid	MMA	At least 5 mL of urine, store and transport frozen	\$100	1 to 4 days. Urgent testing can be performed the same day but must be arranged with the metabolic physician.	Part of metabolic screen
Newborn Screening (NBS)	Newborn Screening (NBS)	Cystic Fibrosis (CF) - Screening test, Guthrie card or Guthrie test, Heel Prick, Hypothyroidism (CH), IRT, MCAD Screening, MSMS, Phenylalanine, PKU Screening, Tandem mass spec screening, TSH	Newborn screening (Guthrie) card, available from the laboratory. Air-dry the blood spots for at least 4 hours without heat before transport to the laboratory. <u>Do not place or transport in plastic bags.</u>	No charge	Next day	

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