




	<h2 style="margin: 0;">MITOCHONDRIAL LABORATORY</h2>		 Murdoch Childrens Research Institute <i>Healthier Kids. Healthier Future.</i>
Test	Specimen Requirements	Cost	Turnaround time
Respiratory Chain Enzymes	Preferred sample: Skeletal muscle, liver, cardiac muscle (depending on clinical history).	\$1,075	6 weeks
Synonyms *Cytochrome c oxidase *Cytochrome oxidase *Electron Transport Chain (ETC) *Mitochondrial - enzyme analysis *Oxidative Phosphorylation *Oxphos *RC enzymes	Skin fibroblast & transformed lymphoblast cell lines are suitable only in certain circumstances. Tissue biopsy/autopsy samples must be stored at -70°C (-20°C is inadequate) until transport on dry ice. Once frozen, they must not be allowed to thaw.	\$1,450	6 months
	Postmortem Samples: COLLECT AS SOON AFTER DEATH AS POSSIBLE , preferably within 2 hours. Note the time between death and freezing of samples. Tissue samples should be no larger than 1.0 cm cubes; heart, kidney & brain samples may also be collected. Enzymes can be assayed in postmortem heart but endomyocardial microbiopsies are too small. Cultured cells should be sent at room temperature and must not be allowed to freeze.		
	OTHER INFORMATION NEEDED FOR RELIABLE INTERPRETATION:		
	1. Patient name, medical record no., date of birth, date of sample, delay between death and freezing if relevant		
	2. Paired plasma lactate & pyruvate levels; multiple estimations preferred.		
	3. Paired CSF lactate & pyruvate levels, preferably with blood levels obtained at about the same time.		
	4. Full clinical summary and family history with pedigree.		
	5. Copies of relevant investigations, as appropriate, including:		
	*urine organic & amino acids		
	*tissue histology (including electron microscopy & enzyme histochemistry)		
	*summary of CT, MRI or MRS studies of brain.		

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Test	Specimen Requirements	Cost	Turnaround time
Pyruvate Dehydrogenase Complex	Preferred sample: 1 flask of skin fibroblasts at an early passage number.	\$690	6 months
Synonyms *PDH - enzyme analysis *PDHC *Pyruvate Decarboxylase *Pyruvate Dehydrogenase - enzyme analysis	Cultured cells should be sent at room temperature and must not be allowed to freeze in transit.		
	Information needed for reliable interpretation:		
	1. Patient name, UR, date of birth, date of sample.		
	2. Paired plasma lactate & pyruvate levels; multiple estimations preferred.		
	3. Paired CSF lactate & pyruvate levels, preferably with blood levels obtained at about the same time.		
	4. Full clinical summary and family history with pedigree.		
	5. Copies of relevant investigations, as appropriate, including:		
	*urine organic & amino acids		
	*tissue histology (including electron microscopy & enzyme histochemistry)		
	*summary of CT, MRI or MRS studies of brain.		
Fumarase	Preferred sample: 1 flask of skin fibroblasts at an early passage number.	\$690	6 months
Synonym *Fumarate Hydratase	Cultured cells should be sent at room temperature and must not be allowed to freeze in transit.		
	Information needed for reliable interpretation:		
	1. Patient name, UR, date of birth, date of sample.		
	2. Full clinical summary, family history and copies of relevant investigations, including uring organic and amino acids.		

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Test	Specimen Requirements	Cost	Turnaround time		
Holocarboxylase Synthetase	Preferred sample: 1 flask of skin fibroblasts at an early passage number	\$690	6 months		
Synonyms *Multiple Carboxylase *Pyruvate Carboxylase *Propionyl CoA Carboxylase	Cultured cells should be sent at room temperature and must not be allowed to freeze in transit.				
	Information needed for reliable interpretation:				
	1. Patient name, UR, date of birth, date of sample.				
	2. Full clinical summary, family history and copies of relevant investigations, including uring organic and amino acids.				
Enzymatic Prenatal Diagnosis	Prenatal diagnosis can only be offered to a subset of families with enzyme defects. Each patient must be discussed in advance.	\$2,500	5 weeks		
	Preferred sample: Chorionic villi dissected free of maternal tissue				
	Chorionic villi in Tissue Culture medium should be sent at room temperature and must not be allowed to freeze in transit.				