

<b>TEST NAME</b>	<b>Pyruvate Dehydrogenase Complex (Lymphocytes)</b>
<b>TEST CODE</b>	PDCL
<b>LOINC</b>	
<b>CPT</b>	82658x2, 82657, 84157
<b>PANEL COMPONENTS</b>	PDC activated, PDC inactivated, Dihydrolipoamide dehydrogenase (E3), PDC:E3 ratio, & Protein
<b>SYNONYMS</b>	PDH, PDHC, PDC, PDHA1
<b>TEST INDICATION</b>	Pyruvate dehydrogenase complex deficiency, typically associated with post-prandial lactic acidemia with a normal lactate/pyruvate ratio, can be assayed in freshly isolated blood lymphocytes, cultured skin fibroblasts, or frozen tissues. The most common defects affect the X-linked E1-alpha subunit, which may be variably expressed especially in females. Measurement in more than one cell type or tissue is recommended because of variable cell/tissue expression with males and females.
<b>METHOD DESCRIPTION</b>	Radioactive enzyme assay measures the rate of decarboxylation of 1-14C-pyruvate, dependent on the presence of thiamine pyrophosphate and coenzyme A, after preactivation or inactivation (by dephosphorylation or phosphorylation) of the enzyme. Assay of the E3 component (dihydrolipoamide dehydrogenase) is used as an internal control for mitochondrial content.
<b>COLLECT</b>	REQUIRES ADVANCE SCHEDULING
<b>SPECIMEN REQUIREMENTS</b>	<p>REQUIRES ADVANCE SCHEDULING</p> <p>*Amt Required varies with patient age (see below).</p> <p>*20ml of control blood from an unrelated adult volunteer, drawn at the same time and in the same way as the patient is also REQUIRED!</p> <p>*Collect blood in ACD (A or B) or CPD-A1 using sterile technique.</p> <p>*ACD is available in yellow stoppered Vacutainer tubes.</p> <p>*NOTE: Yellow stoppered Vacutainer tubes containing SPS, for use in microbiology, are not suitable for enzyme assays.</p>
<b>SAMPLE</b>	Whole Blood
<b>CONTAINER</b>	ACD (A or B) yellow stopper tube
<b>PREFERRED VOLUME</b>	<p>5 ml - patients &lt;6 months old</p> <p>10 ml - patients 6 months - 6 years old</p> <p>20 ml - patients &gt;6 years old</p> <p>**20 ml from an unrelated adult control</p>
<b>MINIMUM VOLUME</b>	None.
<b>TRANSPORT</b>	REQUIRES ADVANCE SCHEDULING
<b>SAMPLE</b>	Whole Blood
<b>PREFERRED VOLUME</b>	<p>5 ml - patients &lt;6 months old</p> <p>10 ml - patients 6 months - 6 years old</p> <p>20 ml - patients &gt;6 years old</p> <p>**20 ml from an unrelated adult control</p>
<b>MINIMUM VOLUME</b>	None.
<b>TEMPERATURE</b>	Room temperature.

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<b>HANDLING</b>	<p>Sterile technique is used to collect blood from the patient and a volunteer unrelated adult control in ACD. Do not freeze or refrigerate. Do not centrifuge. Upon receipt, the blood is stored at room temperature until the lymphocytes are isolated (48-72 hours after blood is obtained).</p> <p>Collection and shipment of whole blood for lymphocyte assays is arranged in advance by calling the CIDEM laboratory. Lymphocytes must be isolated within 48-72 hours of collection, and prior arrangements are needed to reserve sufficient laboratory time to complete the assays.</p>
<b>SHIPPING INSTRUCTIONS</b>	Ship at room temperature.
<b>SPECIAL INSTRUCTIONS</b>	Ship overnight. Prior scheduling required. A 20 ml control from an unrelated is required along with the sample.
<b>PERFORMED</b>	UHCMC; Mon - Fri
<b>TURN AROUND TIME</b>	2 weeks

<b>TEST NAME</b>	<b>Pyruvate Dehydrogenase Complex (Skin Fibroblast)</b>
<b>TEST CODE</b>	PDCFB
<b>LOINC</b>	74577-8
<b>CPT</b>	82658x2, 82657, 84157, 88233
<b>PANEL COMPONENTS</b>	PDC activated, PDC inactivated, Dihydrolipoamide dehydrogenase (E3), PDC:E3 ratio, & Protein
<b>SYNONYMS</b>	PDH, PDHC, PDC, PDHA1
<b>TEST INDICATION</b>	Pyruvate dehydrogenase complex deficiency, typically associated with post-prandial lactic acidemia with a normal lactate/pyruvate ratio, can be assayed in freshly isolated blood lymphocytes, cultured skin fibroblasts, or frozen tissues. The most common defects affect the X-linked E1-alpha subunit, which may be variably expressed especially in females. Measurement in more than one cell type or tissue is recommended because of variable cell/tissue expression with males and females.
<b>METHOD DESCRIPTION</b>	Radioactive enzyme assay measures the rate of decarboxylation of 1-14C-pyruvate, dependent on the presence of thiamine pyrophosphate and coenzyme A, after preactivation or inactivation (by dephosphorylation or phosphorylation) of the enzyme. Assay of the E3 component (dihydrolipoamide dehydrogenase) is used as an internal control for mitochondrial content.
<b>COLLECT</b>	REQUIRES ADVANCE SCHEDULING
<b>SPECIMEN REQUIREMENTS</b>	<p>REQUIRES ADVANCE SCHEDULING</p> <p>Biopsy must be obtained under sterile conditions. Be careful to remove all betadine and alcohol and rinse with saline prior to obtaining the sample. Use a 3 or 4 mm punch biopsy kit or take from the site of incision for a muscle biopsy. Place biopsy in a container of sterile Hanks Balance Salt Solution (HBSS), sterile Ringer's Lactate, or other physiological solution.</p> <p>OR</p> <p>Two confluent T25 flasks filled to the neck with media in a non-vented flask and tape/parafilm shut. If several assays are ordered (more than 3), the shipment of an additional T25 flask is recommended.</p> <p>Also the cell line MUST be MYCOPLASMA tested. If your institution is unable to test for mycoplasma, please tell us when you schedule a shipment.</p>
<b>SAMPLE</b>	Skin biopsy or cultured skin fibroblasts
<b>CONTAINER</b>	A punch biopsy placed into a sterile container with Hanks Balance Salt Solution or physiological saline. OR 2 T25 flasks of confluent cultured fibroblasts.
<b>PREFERRED VOLUME</b>	1 punch biopsy OR 2 T25 flasks of confluent cultured fibroblasts.
<b>MINIMUM VOLUME</b>	None.
<b>TRANSPORT</b>	REQUIRES ADVANCE SCHEDULING
<b>SAMPLE</b>	Skin biopsy OR cultured skin fibroblasts
<b>PREFERRED VOLUME</b>	1 punch biopsy OR 2 T25 flasks of confluent cultured fibroblasts.
<b>MINIMUM VOLUME</b>	None.
<b>TEMPERATURE</b>	Room temperature

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<b>HANDLING</b>	Two confluent T25 flasks filled to the neck with media with a non-vented flask and tape/parafilm shut. If more than three assays are ordered, an additional T25 flask is required.
<b>SHIPPING INSTRUCTIONS</b>	Ship at room temperature
<b>SPECIAL INSTRUCTIONS</b>	Advanced scheduling is required. Ship overnight
<b>PERFORMED</b>	UHCMC; Mon - Fri
<b>TURN AROUND TIME</b>	3-4 weeks for fibroblasts from T25 7-10 weeks from fibroblasts from skin biopsy

<b>TEST NAME</b>	<b>Pyruvate Dehydrogenase Complex (Tissue)</b>
<b>TEST CODE</b>	PDCT
<b>LOINC</b>	74578-6
<b>CPT</b>	82658x2, 82657, 84157
<b>PANEL COMPONENTS</b>	PDC activated, PDC inactivated, Dihydrolipoamide dehydrogenase (E3), PDC:E3 ratio, & Protein
<b>SYNONYMS</b>	PDH, PDHC, PDC, PDHA1
<b>TEST INDICATION</b>	Pyruvate dehydrogenase complex deficiency, typically associated with post-prandial lactic acidemia with a normal lactate/pyruvate ratio, can be assayed in freshly isolated blood lymphocytes, cultured skin fibroblasts, or frozen tissues. The most common defects affect the X-linked E1-alpha subunit, which may be variably expressed especially in females. Measurement in more than one cell type or tissue is recommended because of variable cell/tissue expression with males and females.
<b>METHOD DESCRIPTION</b>	Radioactive enzyme assay measures the rate of decarboxylation of 1-14C-pyruvate, dependent on the presence of thiamine pyrophosphate and coenzyme A, after preactivation or inactivation (by dephosphorylation or phosphorylation) of the enzyme. Assay of the E3 component (dihydrolipoamide dehydrogenase) is used as an internal control for mitochondrial content.
<b>COLLECT</b>	
SAMPLE REQUIREMENTS	After a biopsy/autopsy, the tissue specimen should be immediately quick-frozen in liquid nitrogen.
SAMPLE	Tissue (Muscle, heart, or liver)
CONTAINER	Plastic tube or container
PREFERRED VOLUME	100 mg
MINIMUM VOLUME	50 mg
<b>TRANSPORT</b>	
SAMPLE	Tissue (Muscle, heart, or liver)
PREFERRED VOLUME	100 mg
MINIMUM VOLUME	50 mg
TEMPERATURE	Frozen on dry ice
<b>HANDLING</b>	<p>Tissues obtained at biopsy or autopsy should be immediately frozen in liquid nitrogen or dry ice, and then stored at -60°C until shipment to CIDEM. Tissues must be packed in dry ice and shipped overnight. Samples received at CIDEM are stored at -60°C.</p> <p>Samples, which have thawed during shipment due to insufficient dry ice, or samples which have not been quick frozen or stored at -60°C may produce unsatisfactory results due to deterioration. Samples obtained at autopsy done more than ~8 hours after death may also produce unsatisfactory results due to autolysis. Submitters of such samples will be informed of the uncertainty of obtaining adequate results prior to performing the assay.</p>
<b>SHIPPING INSTRUCTIONS</b>	Ship on dry ice
<b>SPECIAL INSTRUCTIONS</b>	Ship overnight
<b>PERFORMED</b>	UHCMC; Mon - Fri
<b>TURN AROUND TIME</b>	2 weeks