

<b>TEST NAME</b>	<b>Pyruvate carboxylase (Skin Fibroblast)</b>
<b>TEST CODE</b>	PCFB
<b>LOINC</b>	
<b>CPT</b>	82658, 82657, 84157, 88233
<b>PANEL COMPONENTS</b>	PC, Citrate synthase (CS), PC:CS ratio, & Protein
<b>SYNONYMS</b>	PC
<b>TEST INDICATION</b>	Pyruvate carboxylase deficiency, associated with hypoglycemia with severe lactic acidosis, hyperammonemia, ketosis, hepatomegaly, hypomyelination, and difficulty in management, can be assayed in cultured skin fibroblasts, or frozen liver.
<b>METHOD DESCRIPTION</b>	Radioactive enzyme assay of pyruvate dependent fixation of <sup>14</sup> CO <sub>2</sub> following conversion of the oxalacetate product to citrate in the presence of excess citrate synthase and acetyl CoA. Linearity is established with respect to time and amount of sample (protein). Assay of citrate synthase (CS) is used as an internal control for mitochondrial content.
<b>COLLECT</b>	
<b>SPECIMEN REQUIREMENTS</b>	<p><b>REQUIRES ADVANCE SCHEDULING</b></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 <b>MUST</b> 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>	
<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
<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 carboxylase (Liver)</b>
<b>TEST CODE</b>	PCL
<b>LOINC</b>	
<b>CPT</b>	82658, 82657, 84157
<b>PANEL COMPONENTS</b>	PC, Citrate synthase (CS), PC:CS ratio, & Protein
<b>SYNONYMS</b>	PC
<b>TEST INDICATION</b>	Pyruvate carboxylase deficiency, associated with hypoglycemia with severe lactic acidosis, hyperammonemia, ketosis, hepatomegaly, hypomyelination, and difficulty in management, can be assayed in cultured skin fibroblasts, or frozen liver.
<b>METHOD DESCRIPTION</b>	Radioactive enzyme assay of pyruvate dependent fixation of <sup>14</sup> CO <sub>2</sub> following conversion of the oxalacetate product to citrate in the presence of excess citrate synthase and acetyl CoA. Linearity is established with respect to time and amount of sample (protein). Assay of citrate synthase (CS) 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	Liver
CONTAINER	Plastic tube or container
PREFERRED VOLUME	20 mg
MINIMUM VOLUME	5 mg
<b>TRANSPORT</b>	
SAMPLE	Liver
PREFERRED VOLUME	20 mg
MINIMUM VOLUME	5 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