

<b>TEST NAME</b>	<b>Ketoglutarate Dehydrogenase Complex (Lymphocytes)</b>
<b>TEST CODE</b>	KDCL
<b>LOINC</b>	
<b>CPT</b>	82658, 82657, 84157
<b>PANEL COMPONENTS</b>	KDC , Dihydrolipoamide dehydrogenase (E3), PDC:E3 ratio, & Protein
<b>SYNONYMS</b>	KDH, KDHC, KDC
<b>TEST INDICATION</b>	Ketoglutarate dehydrogenase complex deficiency, typically associated with developmental delay and hypotonia, with or without lactic acidemia, can be assayed in freshly isolated blood lymphocytes, cultured skin fibroblasts, or frozen tissues.
<b>METHOD DESCRIPTION</b>	KDC activity is measured by decarboxylation of 1-C14-ketoglutarate, dependent on the presence of thiamine pyrophosphate and coenzyme A. Assay of the E3 component (dihydrolipoamide dehydrogenase) is used as an internal control for mitochondrial content.
<b>COLLECT</b>	
<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>	
<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.

<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>	<p>Ship overnight. Prior scheduling required.</p> <p>A 20 ml control from an unrelated is required along with the sample.</p>
<b>PERFORMED</b>	UHCMC; Mon - Fri
<b>TURN AROUND TIME</b>	2 weeks

<b>TEST NAME</b>	<b>Ketoglutarate Dehydrogenase Complex (Skin Fibroblast)</b>
<b>TEST CODE</b>	KDCFB
<b>LOINC</b>	74577-8
<b>CPT</b>	82658, 82657, 84157, 88233
<b>PANEL COMPONENTS</b>	KDC, Dihydrolipoamide dehydrogenase (E3), KDC:E3 ratio, & Protein
<b>SYNONYMS</b>	KDH, KDHC, KDC
<b>TEST INDICATION</b>	Ketoglutarate dehydrogenase complex deficiency, typically associated with developmental delay and hypotonia, with or without lactic acidemia, can be assayed in freshly isolated blood lymphocytes, cultured skin fibroblasts, or frozen tissues.
<b>METHOD DESCRIPTION</b>	KDC activity is measured by decarboxylation of 1-C14-ketoglutarate, dependent on the presence of thiamine pyrophosphate and coenzyme A. 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
<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>Ketoglutarate Dehydrogenase Complex (Tissue)</b>
<b>TEST CODE</b>	PDCT
<b>LOINC</b>	74578-6
<b>CPT</b>	82658x2, 82657, 84157
<b>PANEL COMPONENTS</b>	KDC, Dihydrolipoamide dehydrogenase (E3), KDC:E3 ratio, & Protein
<b>SYNONYMS</b>	KDH, KDHC, KDC
<b>TEST INDICATION</b>	Ketoglutarate dehydrogenase complex deficiency, typically associated with developmental delay and hypotonia, with or without lactic acidemia, can be assayed in freshly isolated blood lymphocytes, cultured skin fibroblasts, or frozen tissues.
<b>METHOD DESCRIPTION</b>	KDC activity is measured by decarboxylation of 1-C14-ketoglutarate, dependent on the presence of thiamine pyrophosphate and coenzyme A. 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