

TEST NAME	β-Methylcrotonyl-CoA Carboxylase (Skin Fibroblast)
TEST CODE	MCCFB
LOINC	
CPT	82658X2, 82657, 84157, 88233
PANEL COMPONENTS	MCC, Citrate synthase (CS), PC:CS ratio, & Protein
SYNONYMS	MCC
TEST INDICATION	Useful for diagnostic confirmation of specific defects of b-methylcrotonyl-CoA carboxylase (usually recognized through organic acid or acylcarnitine analyses), for distinction from general defects of biotin sufficiency/metabolism, or for detection of heterozygosity. Assay of pyruvate carboxylase (another biotin enzyme) is included as an internal control.
METHOD DESCRIPTION	Radioactive enzyme assay of substrate specific ¹⁴ CO ₂ fixation.
COLLECT	
SPECIMEN REQUIREMENTS	<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>
SAMPLE	Skin biopsy or cultured skin fibroblasts
CONTAINER	A punch biopsy placed into a sterile container with Hanks Balance Salt Solution or physiological saline. OR 2 T25 flasks of confluent cultured fibroblasts.
PREFERRED VOLUME	1 punch biopsy OR 2 T25 flasks of confluent cultured fibroblasts.
MINIMUM VOLUME	None.
TRANSPORT	
SAMPLE	Skin biopsy OR cultured skin fibroblasts
PREFERRED VOLUME	1 punch biopsy OR 2 T25 flasks of confluent cultured fibroblasts.
MINIMUM VOLUME	None.
TEMPERATURE	Room temperature
HANDLING	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.
SHIPPING INSTRUCTIONS	Ship at room temperature
SPECIAL INSTRUCTIONS	Advanced scheduling is required. Ship overnight
PERFORMED	UHCMC; Mon - Fri
TURN AROUND TIME	3-4 weeks for fibroblasts from T25 7-10 weeks from fibroblasts from skin biopsy

TEST NAME	β-Methylcrotonyl-CoA Carboxylase (Liver)
TEST CODE	MCCL
LOINC	
CPT	82658X2, 82657, 84157
PANEL COMPONENTS	MCC, Citrate synthase (CS), PC:CS ratio, & Protein
SYNONYMS	MCC
TEST INDICATION	Useful for diagnostic confirmation of specific defects of b-methylcrotonyl-CoA carboxylase (usually recognized through organic acid or acylcarnitine analyses), for distinction from general defects of biotin sufficiency/metabolism, or for detection of heterozygosity. Assay of pyruvate carboxylase (another biotin enzyme) is included as an internal control.
METHOD DESCRIPTION	Radioactive enzyme assay of substrate specific $^{14}\text{CO}_2$ fixation.
COLLECT	
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
TRANSPORT	
SAMPLE	Liver
PREFERRED VOLUME	20 mg
MINIMUM VOLUME	5 mg
TEMPERATURE	Frozen on dry ice
HANDLING	<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>
SHIPPING INSTRUCTIONS	Ship on dry ice
SPECIAL INSTRUCTIONS	Ship overnight
PERFORMED	UHCMC; Mon - Fri
TURN AROUND TIME	2 weeks