

Test Definition: FETCE

Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts

| _ | | | | |
|---|-----|----|----|---|
| | er/ | VI | e١ | N |

Method Name

Spectrophotometric

NY State Available

Yes

Specimen

Specimen Type

Fibroblasts

Specimen Required

Cultured Fibroblasts

3 T-25 flasks(s) filled to neck with culture media. Maintain sterility and forward promptly at ambient temperature.

Complete and submit with specimen:

1. Baylor Mitochondrial request form.

Reject Due To

| Hemolysis | NA NA |
|-----------|-------|
| Lipemia | NA |
| Icterus | NA |
| Other | NA |

Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Fibroblasts | Ambient | | |

Clinical & Interpretive

Reference Values

A final report will be attached in Mayo Access.

Clinical Reference



Test Definition: FETCE

Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts

Kirby DM, Thorburn DR, Turnbull DM, Taylor RW. Biochemical assays of respiratory chain complex activity. Methods Cell Biol, 2007; 80:93-119.

Trounce, I.A., Kim, Y.L., Jun, A.S., Wallace, D.C. Assessment of mitochondrial oxidative phosphorylation in patients muscle biopsies, lymphoblasts and transmitochondrial cell lines. Methods Enzymol, 1996. 264, 484-509.

Gellerich, F.N. et al. (2004) The problem of interlab variation in methods mitochondrial disease diagnosis: enzymatic measurement of respiratory chain complexes. Mitochondrion 4, (427-439)

Enns, G.M. et al. (2005) Relatonship of primary mitochondrial respiratory chain· dysfunction to fiber type abnormalities in skeletal muscle. Clin. Genet. 66: 337-346. Bernier, FP, Boneh A, Dennett X, Chow CW, Cleary MA, Thorburn DR (2002) diagnostic criteria for respiratory chain disorders in adults and children. Neurology 59, 1406-11.

Walker UA, Collins S, Byrne E (1996) Respiratory chain encephalomyopathies: a diagnostic classification. Eur Neurol 36, 260-267.

Performance

Method Description

The electron transport chain enzymes were assayed at 30 C using a temperature-controlled spectrophotometer. Each assay is performed in duplicate. The activities of complex I (NADH:Ferricyanide dehydrogenase), complex II (succinate-dehydrogenase), complex I+III (NADH:cytochrome c oxidoreductase), complex II+III (succinate:cytochrome c reductase) and complex IV (cytochrome c oxidase) were measured using different electron acceptors/donors. The increase or decrease of cytochrome c at 550 nm was measured for complex I+III, II+III, or complex IV. The activity of complex I was measured-by following the oxidation of NADH at 340 nm. For complex II, the reduction of 2, 6-dichloroindophenol (DCIP) at 600 nm was measured. Citrate synthase is used as a marker for mitochondrial content. Enzyme activities are normalized against citrate synthase (CS) activity when CS activity is greater than 1 standard deviation above or below the control mean. The second figures in parentheses represent data after normalization if it has been performed. The interpretation of the results is done on the assumption that the specimen has been handled properly.

PDF Report

Referral

Day(s) Performed

Upon receipt

Report Available

42 to 45 days

Performing Laboratory Location

Baylor Medical Genetics Laboratories

Fees & Codes

Fees



Test Definition: FETCE

Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts

- Authorized users can sign in to <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.

CPT Code Information

84311 x 6

LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|-----------|----------------------------------|---------------------|
| FETCE | Electron Transport Chain Enzymes | Not Provided |
| | | |
| Result ID | Test Result Name | Result LOINC® Value |
| Z1751 | Electron Transport Chain Enzymes | Not Provided |