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MITOswab Test Results

Specimen ID: **MTP231018**

Name**:** Isaac Franca ***Date of Birth:*** 11/06/2018 ***Dr.*****:** Burch, Mary

MITOswab test: - ***Mitochondrial respiratory chain complexes (RC-I and RC-IV) activities and Citrate Synthase (CS) enzyme activity are measured in patient's buccal cells to evaluate the mitochondrial function in the buccal cells sample.***

Result Values- (Observations)

Activity name	Value *	^Normal Range ^(mean ±SD)
Total Buccal Protein yield (micrograms)	703	
Citrate Synthase[§]	29.24 (242%)	4.4 – 22 (**12.1** ±5.1)
RC-IV (RC-IV/CS)[¶]	0.140 (45%)	0.15 -- 0.6 (**0.31** ±0.1)
RC-I (RC-I/CS)[¶]	3.6 (54%)	3.4 -- 11.9 (**6.8** ±2.0)

Notes-

§: Activity value as nanomoles/min/mg buccal protein

¶: Presented as ratio of the corresponding RC activity to CS activity

*: Number in parenthesis indicates the percent of control mean activity.

^: Based on published data.

Comments- (Explanations)

MITOswab test analysis reveals -

-The overall content of mitochondria was significantly above the normal range as indicated by the citrate synthase activity value (242% **of the normal mean activity level) in test buccal sample.**

- The activity of Respiratory Chain Complex-IV (RC-IV) (45%**of the normal mean value) was below the normal range.**

- The activity of Respiratory Chain Complex-I (RC-I) (54% **of the normal mean value) was in the normal range.**

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Interpretation-

- Biochemical analysis results of subject's buccal sample suggest that it has marginally decreased activity of RC-IV and normal range activity of RC-I, but CS activity was increased significantly above the normal range.

- Almost 2.5-fold higher (than Normal mean value) CS activity may suggest a compensatory function in response to the mitochondrial dysfunction that may be present due to decreased activities of RC-IV.

- The RC-IV deficiency found in the test buccal sample was statistically non-significant.

- Periodic monitoring of mitochondrial enzyme assessment is strongly suggested.

***Notes:* - It is important to note that the buccal mitochondrial enzyme testing approach is still a work in continued improvisations. While the published data supports the evidence for the representation of skeletal muscle (skm) RC enzymes activities by buccal tissue (as found in 84% of cases studied), more work is needed to be undertaken to have a more conclusive statement. Similarly, in brain and neurologic cells, although could not be directly verified, strong correlation has been experienced between buccal cell mitochondrial analysis and some brain or neurological system related problems.**

In addition, the proportion of (ratio) of defective to normal mitochondria, which varies over a broad range, determines the presence and severity of the disease. Thus an accurate assessment of mitochondrial dysfunction might require more than once and independent testing, which is difficult with muscle biopsy and in fact conveniently and easily possible with buccal testing methods. It is recommended to repeat the buccal testing within six months to see if the deficiency is repeatable.

***Disclaimer – Please note that this MITOswab test is a lab developed test (LDT) and not yet approved by FDA*.**

Signed by-

1) Performed and Reviewed by Shirish Damle **Sign and Date** 08/01/2023

2) Medical Director Dr. Jagadish Ulloor

Additional Information:-

The activity assay for mitochondrial respiratory chain complexes **RC-II** and **RC-II+III** were performed.

Activity name	**Value ***	**^ Normal Range ^(mean ±SD)**
RC-II (activity/CS) ¶	0.121 (63%)	0.03 -- 0.35 (**0.194** ±0.08)
RC-II+III (activity/CS)¶	0.018 (19%)	0.032 – 0.152 (**0.092** ±0.03)

*: Number in parenthesis indicates the percent of control mean activity.

¶: Presented as ratio of the corresponding RC activity to CS activity

^: Based on published data.

The results are as follows-

The RC-II activity was **in the normal** range.

The RC-II+III activity was **below the normal** range.

Notes: **- It is important to note that the buccal mitochondrial enzyme testing approach is still a work in continued improvisations. While the published data supports the evidence for the representation of skeletal muscle (skm) RC enzymes activities by buccal tissue (as found in 84% of cases studied), more work is needed to be undertaken to have a more conclusive statement. Similarly, in brain and neurologic cells, although could not be directly verified, strong correlation has been experienced between buccal cell mitochondrial analysis and some brain or neurological system related problems.**

In addition, the proportion of (ratio) of defective to normal mitochondria, which varies over a broad range, determines the presence and severity of the disease. Thus an accurate assessment of mitochondrial dysfunction might require more than once and independent testing, which is difficult with muscle biopsy and in fact conveniently and easily possible with buccal testing methods. It is recommended to repeat the buccal testing within six months to see if the deficiency is repeatable.

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***The RC-I, RC*-II and RC-II+III activity assays validation is not complete and hence the results should be considered as for research purpose only.**

1) Performed and Reviewed by Shirish Damle **Sign and Date** 08/01/2023

2) Medical Director Dr. Jagadish Ulloor