

Accepted Manuscript

Metabolic studies of a patient harbouring a novel S487L mutation in the catalytic subunit of AMPK

Juliana Harumi Arita, Mário H. Barros, Felipe Gustavo Ravagnani, Marcello Ziosi, Lívia Rentas Sanches, Fabíola Rosa Picosse, Tania Oliveira Lopes, Patrícia de Carvalho Aguiar, Carolina Habermann Macabelli, Marcos Chiaratti, José Luiz Pedroso, Catarina M. Quinzi, Orlando Graziani Póvoas Barsottini, Claudia Cristina Ferreiro-Barros



PII: S0925-4439(18)30091-7
DOI: doi:[10.1016/j.bbadis.2018.03.011](https://doi.org/10.1016/j.bbadis.2018.03.011)
Reference: BBADIS 65082

To appear in:

Received date: 28 August 2017
Revised date: 18 February 2018
Accepted date: 7 March 2018

Please cite this article as: Juliana Harumi Arita, Mário H. Barros, Felipe Gustavo Ravagnani, Marcello Ziosi, Lívia Rentas Sanches, Fabíola Rosa Picosse, Tania Oliveira Lopes, Patrícia de Carvalho Aguiar, Carolina Habermann Macabelli, Marcos Chiaratti, José Luiz Pedroso, Catarina M. Quinzi, Orlando Graziani Póvoas Barsottini, Claudia Cristina Ferreiro-Barros, Metabolic studies of a patient harbouring a novel S487L mutation in the catalytic subunit of AMPK. The address for the corresponding author was captured as affiliation for all authors. Please check if appropriate. *Bbadis*(2018), doi:[10.1016/j.bbadis.2018.03.011](https://doi.org/10.1016/j.bbadis.2018.03.011)

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

Metabolic studies of a patient harbouring a novel S487L mutation in the catalytic subunit of AMPK.

Juliana Harumi Arita^{1*}; Mário H Barros^{2*}; Felipe Gustavo Ravagnani³; Marcello Ziosi⁸; Lívia Rentas Sanches⁴; Fabíola Rosa Picosse⁵; Tania Oliveira Lopes⁴; Patrícia de Carvalho Aguiar^{4,6}; Carolina Habermann Macabelli⁷; Marcos Chiaratti⁷; José Luiz Pedroso⁶; Catarina M. Quinzi⁸; Orlando Graziani Póvoas Barsottini⁶ and Claudia Cristina Ferreiro-Barros⁹.

1 – Setor de Neurologia Infantil, Departamento de Neurologia, Universidade Federal de São Paulo – São Paulo, SP, Brazil;

2 – Instituto de Ciências Biomédicas, Universidade de São Paulo – São Paulo, SP, Brazil;

3 – Instituto de Química, Universidade de São Paulo – São Paulo, SP, Brazil;

4 – Hospital Israelita Albert Einstein – São Paulo, SP, Brazil;

5 – Departamento de Dermatologia, Universidade Federal de São Paulo – São Paulo, SP, Brazil;

6 – Departamento de Neurologia e Neurocirurgia, Universidade Federal de São Paulo – São Paulo, SP, Brazil;

7 – Departamento de Genética e Evolução, Universidade Federal de São Carlos, UFSCar, São Paulo, SP, Brazil;

8 – Department of Neurology, Columbia University Medical Center – New York, NY, USA;

9 – Departamento de Clínica Médica, Faculdade de Medicina da Universidade de São Paulo – São Paulo, SP, Brazil.

*** Contributed equally for this work.**

Running title: *PRKAA1* S487L homozygous mutation in a patient with neurological impairment treated with exogenous CoQ10.

Corresponding author: Claudia C. Ferreiro-Barros, Departamento de Clínica Médica, Faculdade de Medicina da Universidade de São Paulo – Av. Dr. Arnaldo, 455, São Paulo – SP, Brazil. CEP: 01246-000. Phone number: +55-11-30618249. E-mail: claucfb@gmail.com

Download English Version:

<https://daneshyari.com/en/article/8258491>

Download Persian Version:

<https://daneshyari.com/article/8258491>

[Daneshyari.com](https://daneshyari.com)