Genetics and Molecular Research

Respiratory chain complex III deficiency in patients with *tRNA-leu* mutation

J. Jiang[1], X.L. Wang[2] and Y.Y. Ma[3,4]

[1]Department of Oncology, Affiliated Hospital of Qinghai University, Xining, China
[2]Operation Room, Affiliated Hospital of Qinghai University, Xining, China
[3]Department of Pediatrics, Chinese Liberation Army General Hospital, Beijing, China
[4]Department of Pediatrics, Affiliated Hospital of Qinghai University, Xining, China

Corresponding author: Y.Y. Ma
E-mail: xnmayanyan@126.com

Genet. Mol. Res. 14 (4): 18629-18636 (2015)
Received August 25, 2015
Accepted October 25, 2015
Published December 28, 2015
DOI http://dx.doi.org/10.4238/2015.December.28.12

ABSTRACT. The aim of this study was to investigate the clinical and genetic profiles of mitochondrial disease resulting from deficiencies in the respiratory chain complex III. Three patients, aged between 8 months and 12 years, were recruited for this study. The activities of mitochondrial respiratory chain complexes in the peripheral leucocytes were spectrophotometrically measured. The entire mitochondrial DNA (mtDNA) sequence was analyzed. Samples obtained from the three patients and their families were subjected to restriction fragment length polymorphism and gene sequencing analyses. mtDNA copy numbers of all patients and their mothers were analyzed. The patients displayed nervous system impairment, including motor and mental developmental delay, hypotonia, and motor regression. Two patients also suffered from Leigh syndrome. Assay of the mitochondrial respiratory chain enzymes revealed an isolated complex III deficiency in the three patients. The m.3243 A>G mutation was detected in all patients and their mothers. The mutation loads were 48.3, 57.2, and 45.5% in the patients, and 20.5, 16.4, and 23.6% in their respective mothers. The leukocyte mtDNA copy numbers of the patients

and their mothers were within the control range. The clinical manifestation and genetics were observed to be very heterogeneous. Patient carrying an m.3243 A>G mutation may biochemically display a deficiency in the mitochondrial respiratory chain complex III.

Key words: Respiratory chain complex III deficiency; Mitochondrial DNA; 3243A>G mutation

