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Prof. Dr. K. WIDHALM  
University of Vienna, Department of Pediatrics  
Währinger Gürtel 18-20, A-1090 Vienna

ABSTRACT FORM

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PRESYMPTOMATIC DIAGNOSIS OF HYPOPARATHYROIDISM IN PATIENTS WITH MITOCHONDRIAL DISEASE. Gabriela M. Repetto<sup>1</sup>, Ingrid A. Holm<sup>1,2</sup>, Frances E. Dougherty<sup>1</sup>, Division of <sup>1</sup>Genetics and <sup>2</sup>Endocrinology, Children's Hospital, Boston, USA.

Mitochondrial diseases have wide phenotypic expression due to the ubiquitous nature of the oxidative phosphorylation pathway. Hypoparathyroidism is a known, but rarely reported complication. Two patients with known mitochondrial diseases were found to have low parathyroid hormone (PTH) levels during routine multisystem surveillance, consistent with the diagnosis of hypoparathyroidism. Neither patient showed classic signs and symptoms of hypoparathyroidism. Patient 1, a 14 year old girl with Kearns-Sayre syndrome diagnosed at 8 years of age, had ophthalmoplegia, ptosis, heart block, short stature and increased CSF protein. She complained of weakness and fatigue of 3-4 months duration. She had serum calcium level of 6.9 mg/dl (normal 8.5-9.5), phosphate of 6.9 mg/dl (normal 2.7-4.5), PTH <10 pg/ml (normal >15) with normal vitamin D and magnesium levels. Patient 2 is an 8 year old boy with complex I deficiency diagnosed at 5 years, who presented with developmental delay, myopathy, seizures, lactic acidosis and pancreatic insufficiency. He had normal calcium and phosphate levels but an inappropriately low PTH of <10 pg/ml. Patient 1 began treatment with calcium and vitamin D, resulting in normalization of her calcium and phosphate levels and in improvement of her fatigue. Patient 2 is being monitored without treatment. This report emphasizes 1) the need for periodic multisystem surveillance in patients with mitochondrial diseases for presymptomatic detection of complications, 2) the relevance of including PTH levels in screening these patients, and 3) that hypoparathyroidism is a treatable complication of mitochondrial diseases. Patients with mitochondrial myopathies may be particularly susceptible to muscular manifestations of abnormal calcium regulation, thereby making systematic screening even more crucial.

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AUTHOR'S CORRESPONDENCE ADDRESS:

LASTNAME REPETTO FIRSTNAME GABRIELA TITLE MD SEX M  F

ADDRESS CHILDREN'S HOSPITAL - ENDERS 5 - 300 LONGWOOD AVENUE

POSTAL/ZIP CODE 02115 CITY BOSTON, MA COUNTRY USA

TELEPHONE 1-617-355-8046 FAX 1-617-355-7588

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