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SHORT REPORT

Barth syndrome presenting with acute metabolic decompensation in the neonatal period

Maria Alice Donati · Sabrina Malvagia · Elisabetta Pasquini · Amelia Morrone · Giancarlo La Marca · Barbara Garavaglia · Daniela Toniolo · Enrico Zammarchi

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Summary We describe two patients affected by Barth syndrome. Their symptoms became manifest on respectively the third and first day of their lives. Clinical presentation included poor sucking, lethargy, hypotonia, hypothermia and cardiomyopathy. Laboratory findings such as hypoglycaemia, metabolic acidosis, elevated transaminases, hyperlactacidaemia and mild hyperammonaemia pointed to an inborn error of energy metabolism with possible mitochondrial involvement. Molecular analysis of the *TAZ* (*G4.5*) gene showed the c.877G > A mutation leading to the G197R amino acid substitution in patient 1, and the new splice donor c.829 + 1G > A genetic lesion in patient 2.

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M. A. Donati · S. Malvagia · E. Pasquini · A. Morrone · G. L. Marca · E. Zammarchi (✉)
Metabolic and Muscular Unit, Department of Pediatrics, University of Florence, AOU-Meyer, Florence, Italy; Department of Pediatrics, Meyer Children's Hospital, Via Luca Giordano 13, 50132 Florence, Italy
e-mail: enrico.zammarchi@unifi.it

B. Garavaglia
Molecular Neurogenetics, National Neurological Institute 'Carlo Besta', Milan, Italy

D. Toniolo
Dibit-San Raffaele Scientific Institute, Milan, Italy

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