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SHORT REPORT

Barth syndrome presenting with acute metabolic decompensation in the neonatal period

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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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