# A Case of Mitochondrial Trifunctional Protein Deficiency with Severe Heart Failure of Newborn with *HADHA* Gene Mutation

Takahiko HIRAI [1)], Eiji OHTA [1),2)], Hiroyasu KAWANO [1),2)],
Toshikazu NIIMI [1),2)], Ayako GOTO [1),2)], Takashi SETOUE [1),2)],
Chizuru HASHIGUCHI [1),2)], Masatoshi NAKAMURA [1),2)], Seiji YAMAGUCHI [3)],
Shinichi HIROSE [1),2),4)]

1) *Department of Pediatrics, Faculty of Medicine, Fukuoka University Hospital*
2) *Division of Neonatology, Center for Maternal, Fetal and Neonatal Medicine, Fukuoka University Hospital*
3) *Department of pediatrics, Shimane University*
4) *Department of Pediatrics, Faculty of Medicine, Fukuoka University*

**Abstract**

Mitochondrial trifunctional protein (TFP) deficiency is a fatty acid metabolism disorder that is targeted in neonatal tandem mass screening. When an infant has a TFP deficiency, the mother may also have severe liver dysfunction (e.g., HELLP syndrome or acute fatty liver of pregnancy). Although early detection can be preventive, severe cases manifest in neonates in the form of serious cardiomyopathy, and there is an increased risk of death. We treated a neonate who developed acute heart failure at the age of 2 days. According to blood test results during hospitalization, her mother exhibited liver function impairment. The results of tandem mass screening after birth suspected TFP deficiency, and subsequent genetic testing of the parents and infant resulted in a definitive diagnosis. The infant's cardiac function rapidly declined, and treatment resulted in little improvement. She passed away at the age of 44 days. Genetic evaluation indicated that the infant had a mutation in *HADHA* [c.361C>T (p.O121*), IVS16+2T>G], which encodes TFP. Moreover, both the parents were carriers of this gene. Thus, if an infant delivered by a mother with severe liver function impairment exhibits acute heart failure, it is important to immediately perform tandem mass screening to determine the possibility of a TFP deficiency.

**Key words: Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency, Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency, Acylcarnitine profile analysis, bezafibrate**