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Affiliation(s) (Times New Roman, 9 point, left justification [running numbers] your lab, school or institute, and university)

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Abstract

Body of text **(**Times New Roman, 10.5 point, right and left justification; maximum 400 words**).** Please **do not use indents** at the beginning of paragraphs – we will delete the indents if we find these cases. Please **DO NOT** include the word "Abstract" at the top.

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Mutations in the zinc transporter *ZnT2* gene result in zinc deficiency in a breast-fed infant

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Zinc is an essential mineral and has extensive roles in developmental processes. Therefore, zinc deficiency in infants can result in various disorders including growth restriction, skin lesions, alopecia and immune system dysfunctions. Zinc concentrations in breast milk are considerably higher than those of the maternal serum to meet infant's requirements. Thus, effective mechanisms ensuring secretion of large amounts of zinc into the milk operate during lactation in mammary epithelial cells. The zinc transporter ZnT2 and ZnT4 are thought to be involved in transporting zinc into the milk. Recently we found a Japanese mother with low milk zinc concentrations (>90% reduction) whose infant developed severe zinc deficiency. To investigate the cause of the milk zinc deficiency, we isolated the genomic DNA from the mother's blood and sequenced the *ZnT2* and *ZnT4* genes. We found no mutations in the *ZnT4* gene, but identified two novel missense mutations, causing W152R and S296L substitution, on different alleles in the *ZnT2* gene. Next, we characterized these ZnT2 mutants biochemically using zinc-sensitive DT40 cells. The W152R mutant abolished the activity to transport zinc and to form dimer complex, which is required for the ZnT2 to transport zinc. These results indicated the W152R mutant is a loss-of-function. The S296L mutant retained both abilities but was extremely destabilized. Taken together, the compound heterozygous mutations in the *ZnT2* gene of the mother caused low milk zinc concentrations and resulted in severe zinc deficiency in the breast-fed infant. Our results show that ZnT2 doubtlessly plays an essential role in zinc secretion into milk.

Keywords: Zinc transporter, ZnT2, Mutation, Human disease