

PREGNANCY-RELATED RISKS AND COMPLICATIONS IN WOMEN WITH CITRIN DEFICIENCY: LOW RISK OF PERINATAL HYPERAMMONEMIA AND CAUTION WITH OGTT

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Background: In urea cycle disorders (UCDs), perinatal hyperammonemia is a known risk, raising concern among women with citrin deficiency (CD). There is limited evidence on how pregnancy and delivery impact CD, particularly regarding hyperammonemia, hypoglycemia, or impaired glucose tolerance. This review highlights the need for further investigation into pregnancy-related events and complications in women diagnosed with CD, to better understand the risks and provide insights for those considering pregnancy.

Methods: We retrospectively reviewed medical records from pregnancy and childbirth, including basic health information, blood/urine glucose levels, and adverse events, in Japanese women with CD.

Results: Four CD patients who experienced deliveries were identified. Three of them were undiagnosed until at least one of their children was diagnosed with CD, and the other one was diagnosed based on the typical eating habits. A total of 10 deliveries were documented, with maternal ages at delivery as follows: (#1) 23, 34, and 38 years; (#2) 25 and 26 years; (#3) 29, 31, and 33 years; and (#4) 25 and 27 years. All 10 neonates were born at full term, with an average birth weight of 2,679 g (2,479 g for the five CD children and 2,878 g for the five non-CD children). No CD-related clinical adverse episodes, such as hypoglycemia or hyperammonemia, were observed during pregnancy or childbirth. Interestingly, three women who already exhibited the typical eating habits due to CD reported that this tendency became more pronounced during pregnancy compared to their usual non-pregnancy diet. In case #1, mild gestational diabetes mellitus developed during her last pregnancy. She underwent an oral glucose tolerance test (OGTT) at 25 weeks of gestation, resulting in nausea, dizziness, and loss of consciousness after a 50g glucose load. Her blood glucose levels were managed with insulin therapy without complications, and no hypo- or hyperglycemic episodes occurred during pregnancy.

Conclusions: In contrast to other urea cycle disorders, the risk of hyperammonemia during the perinatal period in CD appears to be low, and the mechanism underlying its onset seems distinct. However, OGTT should be approached with extreme caution in cases where gestational diabetes is suspected, as it poses a high risk of exacerbating the patient's condition.