Prenatal Genetics, Reproductive Genetics

AB115. Prenatal diagnosis for severe cases of targets in expanded newborn screening

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Background: Expanded newborn screening for amino and organic acidemias as well as fatty acid oxidation defects (FAODs) using MS/MS (TMS screening) is becoming popular worldwide. Although many of babies detected in the presymptomatic stage by TMS screening can be saved, a small part of patients have extremely severe course with early death. In such cases, only prenatal diagnosis may be an optional measure for the family.

Methods: We have performed the prenatal diagnosis using GC/MS and TMS as well as gene analysis after amniocentesis at around 16 weeks of gestation. The supernatant was used for acylcarnitine analysis by TMS, and organic acid analysis by selected ion monitoring mode of GC/MS with t-butyldimethylsilyl derivatization, and the pellet was used for gene analysis.

Results: In our experience, 50 of 143 cases introduced to our laboratory at Shimane University were affected, with no misjudges to now.

Conclusions: Misdiagnosis must not be acceptable in prenatal diagnosis. For this reason, it may be preferable that prenatal diagnosis is performed by as many kinds of methods as possible like our system. Prenatal diagnosis of organic acidemias is possible if stable isotopes as internal standard are available for organic acid analysis using GC/MS. In case of FAODs, gene analysis will be essential other than mass spectrometric measure.

Keywords: Prenatal diagnosis; expanded newborn screening; organic acidemia; fatty acid oxidation defect (FAOD)

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