Newborn Screening, Inborn Errors of Metabolism

AB101. Improvement in the sensitivity of newborn screening for Fabry disease among females through the use of a high-throughput and cost-effective method, DNA mass spectrometry

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Background: Until July 2016, more than 916,000 newborns have been screened for Fabry disease by our team. From our estimation, around 80% female newborns will be missed by our current enzyme-based screening. However, female carriers of Fabry disease may still develop severe morbidity and mortality, so our team developed a cost-effective screening method for female newborns with Fabry disease.

Methods: In Taiwan, around 98% of Fabry patients were limited to only 21 pathogenic mutations. Therefore, an Agena iPLEX platform was designed by our team to analyze these 21 pathogenic mutations using only one assay panel.

Results: A total of 54,791 female infants were screened: 136 female newborns with IVS4+919G>A mutation and one with c.656T>C mutation were identified by the new screening method. But 83% of these female newborns (113 cases with IVS4+919G>A, and 1 case with 656T>C) will be missed by the current enzyme-based screening method. Through family study of the female newborns with IVS4+919G>A mutation, 30 adult family members with the mutation were found to have left ventricular hypertrophy. Nine patients underwent endomyocardial biopsy which showed Fabry cardiomyopathy, and received enzyme replacement therapy.

Conclusions: We demonstrated that the Agena iPLEX assay is a powerful tool to screen Fabry females. Through this screening, we could identify several undiagnosed adult family members.

Keywords: Fabry disease; IVS4+919G>A; Agena iPLEX assay; newborn screening; GLA genotyping

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