عنوان مقاله:

First Report of Preimplantation Genetic Diagnosis for Steroid-Resistant Nephrotic Syndrome

محل انتشار:

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خلاصه مقاله:

Background: Steroid-resistant nephrotic syndrome is a genetic disease with autosomal recessive inheritance pattern and symptomssuch as proteinuria and hypoalbuminemia and rapid progress of kidney disease. Preimplantation genetic diagnosis is anoption for couples who are at risk of affected pregnancy to have a healthy child.Objectives: This study aimed to develop a new PGD test for a couple who are heterozygous for a mutation in NPHS۲ gene and havea son affected to steroid-resistant nephrotic syndrome.Methods: Variant detection by cycle sequencing and Multiplex fluorescent PCR for identification of flanking STR markers were usedto investigate the status of the embryos.Results: Three out of six embryos were transferable from which one was transferred and resulted in the birth of a healthy boy.Conclusions: We recommend increasing the number of the STR markers to two at the downstream of the NPHS۲ .gene especially incases that direct mutation analysis such as cycle sequencing is not applied

کلمات کلیدی:

Preimplantation Genetic Diagnosis, Nephrotic Syndrome Type ۲, Steroid-Resistant Nephrotic Syndrome