Free Papers - Abstracts

OP 01 - Simple and cost-effective genetic screening tool to genotype multiple polymorphisms of polycystic ovary syndrome (PCOS)

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Introduction: Polycystic ovary syndrome (PCOS) is a multigenic, endocrine disorder of young women. Inheritance of PCOS is likely to be oligogenic and genetic basis remains largely unknown. The genetics of PCOS requires an in-depth study, particularly among Sri Lankan women who have a greater metabolic risk from an early age. Screening the SNPs of PCOS individually is time consuming. Hence, developing a tool that would help in screening SNPs of multiple candidate genes simultaneously is essential to determine the genetic basis of PCOS. This study aimed to develop a simple and cost-effective genetic screening tool to simultaneously genotype 16 SNPs of PCOS.

Methods: Representative samples from previous studies were taken for the genotyping analysis using the developed tool. The tool was developed using allele specific real time quantitative PCR (AS-qPCR) in 96 well PCR plate. Eight SNPs identified in our previous study as well as 8 SNPs identified from other reported studies that had a strong association in the etiology of PCOS were used to develop the tool. Each single run consists of positive control samples, sample of unknown genotype and negative control. Ten samples (10 runs) were genotyped using this tool and 16 SNPs were genotyped simultaneously in a single run.

Results: Genetic screening tool results were validated with Sanger sequencing and all except 3 genotype results agreed with Sanger sequencing. The tool showed 99% specificity and 97% sensitivity.

Conclusion: The developed genetic screening tool has excellent potential in determining the genotype of multiple SNPs of PCOS simultaneously. This tool is highly suitable for developing countries as a cost effective and accurate early genetic screening test for PCOS. Thus, it provides a reliable and fast genotyping method facilitating a wider implication in clinical practice.

Keywords: PCOS, Single nucleotide polymorphism (SNP), Allele specific qPCR

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