8p23 beta-defensin copy number determination by single-locus pseudogene-based paralog ratio tests risk bias due to low-frequency sequence variations.

Zhang X, Müller S, Möller M, Huse K, Taudien S, Book M, Stuber F, Platzer M, Groth M (2014) 8p23 beta-defensin copy number determination by single-locus pseudogene-based paralog ratio tests risk bias due to low-frequency sequence variations. *BMC Genomics* 15, 64-64.

Abstract

The copy number variation (CNV) in beta-defensin genes (DEFB) on human chromosome 8p23 has been proposed to contribute to the phenotypic differences in inflammatory diseases. However, determination of exact DEFB CN is a major challenge in association studies. Quantitative real-time PCR (qPCR), paralog ratio tests (PRT) and multiplex ligation-dependent probe amplification (MLPA) have been extensively used to determine DEFB CN in different laboratories, but inter-method inconsistencies were observed frequently. In this study we asked which one is superior among the three methods for DEFB CN determination.

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