**ABSTRACT INSTRUCTIONS (delete before submitting)**

Head the abstract with the title; authors; and affiliation(s).

Each of these should be on a separate line. **No abstract should exceed 300 words.**

Abstracts should contain a statement of the problem, brief methods, clear results, and a statement of the conclusions or significance of the findings. Abstracts will be reviewed and may be returned to the authors for modification.

**Abstract Title – Bold, font size 14**

**G-quadruplex DNA structures, genomic imprinting and allelic drop-out in PCR**

**Authors – font size 12, with presenting author underlined.**

Stevens, A.J.1, Stuffrein-Roberts, S.1, Miller, A.L.1, Gibb, A.1, Doudney, K.1, Bagshaw, A.1, Aitchison, A.1, Eccles, M.R.2, Filichev, V.V.3, Kennedy, M.A.1

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**Main text – font size 12, justified, single line spacing**

During analysis of the promoter region of the human *MEST* gene, we noted apparent non-Mendelian behaviour of three closely linked single nucleotide polymorphisms (SNPs)[1](file:///C%3A%5CUsers%5Cjade%5CAppData%5CLocal%5CMicrosoft%5CWindows%5CTemporary%20Internet%20Files%5CContent.Outlook%5CG1ED47IK%5CQMBAbstractTemplate2013.doc#_ENREF_1). *MEST* plays a role in mammalian development and maternal behaviour. It is genomically imprinted, with the maternally inherited allele permanently switched off. When we genotyped these SNPs in many subjects, no heterozygotes were observed, despite the use of multiple PCR-based methods and several different primer pairs. Experiments with mixing the genomic DNA from different individuals proved that the assays were capable of detecting both alleles simultaneously. This indicates that the observed homozygosity was likely resulting from consistent allelic dropout of one allele in every subject.

It is possible that the DNA (CpG) methylation likely to occur on the imprinted allele could play a role in altering the outcome of genotyping results; however, this phenomenon alone cannot explain the pattern of allelic dropout. Therefore we examined the region containing the three SNPs for evidence of secondary structures that might also be a factor in allelic dropout. The region is GC-rich, and using several prediction algorithms[2](file:///C%3A%5CUsers%5Cjade%5CAppData%5CLocal%5CMicrosoft%5CWindows%5CTemporary%20Internet%20Files%5CContent.Outlook%5CG1ED47IK%5CQMBAbstractTemplate2013.doc#_ENREF_2), [3](file:///C%3A%5CUsers%5Cjade%5CAppData%5CLocal%5CMicrosoft%5CWindows%5CTemporary%20Internet%20Files%5CContent.Outlook%5CG1ED47IK%5CQMBAbstractTemplate2013.doc#_ENREF_3) it appeared likely that it has a propensity for forming G-quadruplex (G4) structures. These arise from the formation of G-tetrads by hydrogen bonding of four G residues, either within or between strands, and the subsequent stacking of these into higher order structures. We hypothesized that DNA methylation may interact to stabilize such secondary structures and block the *Taq* polymerase from actively replicating one template strand.

**Optional references – font size 10, Calibri. Title in Italics**

1. Stuffrein-Roberts, S., *Allelic expression patterns in psychatric candidate genes. PhD Thesis* in *Pathology*. 2008, University of Otago: Christchurch. p. 216.

2. Kikin, O., L. D'Antonio and P.S. Bagga (2006). *QGRS Mapper: a web-based server for predicting G-quadruplexes in nucleotide sequences.* Nucleic acids research. 34: W676-82.