



Call for Abstracts

Dear Colleagues

For the 24th International Colloquium on Animal Cytogenetics and Genomics (University of Kent 6-8th July 2020) we are now calling for abstracts in (but not restricted to) the following areas:

- “Clinical” cytogenetics (particularly non-human)
- Genome assembly and chromosomes
- Genome and chromosome evolution
- Karyotypic diversity
- Comparative (cyto) genomics
- Comparative genomics
- Meiosis and mitosis
- Sex chromosomes and specialist chromosomes
- Chromatin in gametic and somatic cells
- Epigenetics and development
- New technologies
- Genome organization and architecture
- Chromosomes in reproduction
- Centromeres and telomeres
- Population cytogenetics
- Outreach and education



Please email your abstract to:

icacg2020@kent.ac.uk

Deadline 30th March (best abstracts will be selected for oral presentation)

Please follow these guidelines (an example is below)

- Use Calibri font 11
- Title in bold
- Presenting author first, surname, initials (no commas or full-stops), superscript number to indicate affiliation, comma
- Affiliations in numerical order
- No more than 500 words – 1 optional diagram–1 page only (abstract not conforming may be returned)

Universal approaches for detecting genetic disease in IVF embryos: towards understanding the cytogenetics of early human development

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Preimplantation Genetic Diagnosis (PGD) – the diagnosis of genetic disorders in human IVF embryos - is about to enter its 25th year as clinical procedure. From the outset however, it has had been impeded by the time consuming, expensive delay associated with tailoring a bespoke test to each couple before treatment. The further complication of having to ability only to detect either the monogenic disorder in question or the chromosomal complement of the embryo potentially limits its application. Interrogation of single nucleotide polymorphism microarrays (SNP chips) can facilitate high-resolution cytogenetic diagnosis and recently we adapted this technology for monogenic disorder detection also (Karyomapping - Handyside et al. 2010). By linkage analysis of parental genotypes, an affected sibling and single cells from IVF embryos Karyomapping makes it possible to identify informative loci for each of the four parental haplotypes on each chromosome and map the inheritance pattern of the disease loci on them. In recent months Karyomapping has entered clinical validation, which has resulted in pregnancies and live births. Karyomapping is potentially capable of detecting the spectrum of monogenic and chromosome disorders and thus has the potential for widespread and global application. Moreover it is a tool that can, for the first time, detect the incidence and origin of chromosome abnormalities in early human development. Uniform chromosome abnormalities in human preimplantation embryo at cleavage stages have their origin in meiotic errors in the gametes whereas post-zygotic mitotic errors result in chromosomal mosaicism. While the levels of chromosome abnormality have been described in detail in ongoing pregnancies and live births, studies in preimplantation human embryos are based on limited technologies that do not have the ability to determine the phase and parent of origin. Karyomapping using genome wide SNP analysis can be applied to sperm, oocytes, single blastomeres or whole embryos to determine not only the existence of chromosome errors but also their origin. Evidence suggests that aneuploidy of maternal meiotic origin shows significant differences to those seen in pregnancy, and that post zygotic changes (principally whole chromosome losses) are commonplace. Patterns of nuclear organisation similar to those seen in committed cells can be observed, particularly in aneuploid nuclei.

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