


Programme and Abstract Book

# hgm

# 2010

 HUMAN  
GENOME  
MEETING

Montpellier, France  
18 - 21 May 2010

**The HUGO Journal**  
Volume 4 Supplement 1 2010



Human Genome Organisation



To achieve that, we set out to establish a technology platform that would allow enrichment for the cells of interest by merging the technologies available from the genomics field with classical molecular genetic approaches using mouse as a model.

A developmental gene with a known expression pattern was engineered endogenously with the enhanced green fluorescent protein (EGFP) by homologous recombination in mouse embryonic stem cells (mESC). The modified mESC were microinjected to generate a transgenic line of mice. The expected EGFP expression pattern was observed in heterozygous embryos at midgestation, and the cell populations expressing the gene of interest were efficiently isolated by Fluorescent Activated Cell Sorting (FACS) to greater than 90% purity. RNA was extracted from the sorted cells and subsequent expression profiling studies done using Illumina Mouse WG-6 Microarrays.

From our work, we have determined the smallest number of cells required for us to obtain the minimum RNA necessary for reproducible gene expression profiling on the Illumina microarray platform. By comparing the expression profiles of EGFP+ and EGFP- cells, we identified a list of genes, including novel and known genes, which are highly expressed in the specific cell lineage isolated.

In summary, a robust strategy to isolate low abundance cell population to high purity for the analysis of spatio-temporal changes in the transcriptome of any specific developing tissue or organ in vivo has been established.

**Poster No: P084-W**

**From HGM10 (1989) to HGM 2010: IG and TR gene concept and IMGT/GENE-DB**

<sup>1</sup>Véronique Giudicelli and <sup>1</sup>Marie-Paule Lefranc

<sup>1</sup>IMGT®, the international ImMunoGeneTics information system®, Laboratoire d'ImmunoGénétique Moléculaire LIGM, Université Montpellier 2, Institut de Génétique Humaine IGH, UPR CNRS 1142, 141 rue de la Cardonille, 34396 Montpellier cedex 5, France

In 1989, at HGM10 in New Haven (USA), the T cell receptor gamma genes were, for the first time, entered in the newly created Genome DataBase (GDB). This was a first major step as it acknowledged the concept of gene for the antigen receptors, the immunoglobulins (IG) and T cell receptors (TR), despite their unique particularities. Indeed, IG and TR chains are coded by genes belonging to four types, variable (V), diversity (D), joining (J) and constant (C), which show an unusual molecular organization due to the necessity of DNA rearrangements for the IG and TR chain synthesis, in B and T cells, respectively. These rearrangements contribute to the huge diversity and fine specificity of the variable domains of the IG and TR that bind specifically to the antigens, in the adaptive immune response. The potential expressed repertoire is estimated to 2x10 to the twelfth different IG and TR per individual. Owing to these particularities, IMGT®, the international ImMunoGeneTics information system® (<http://www.imgt.org>), was created in 1989 by Marie-Paule Lefranc at LIGM, Montpellier, France. IMGT® is a high-quality integrated knowledge resource specialized in the IG, TR, major histocompatibility complex (MHC), immunoglobulin superfamily (IgSF), MHC superfamily (MhcSF) and related proteins of the immune system (RPI) of human and other vertebrate species. IMGT/GENE-DB (Nucl. Acids Res., 33, D256-D261, 2005), the IMGT® genome database, was developed to standardize and classify the IG and TR gene data and to manage the related knowledge. The official nomenclature of human IG and TR genes and alleles, based on IMGT-ONTOLOGY, the first ontology for immunogenetics, was approved in 1999 by the HUGO Nomenclature Committee (HGNC) and acknowledged by the WHO-IUIS. Rules for the identification of gene and allele functionality were defined, with each IG and TR gene and allele being represented by an IMGT reference sequence. Another breakthrough is the IMGT unique numbering and its graphical representation, the IMGT Collier de Perles, which allow the standardization per domain type. In March 2010, IMGT/GENE-DB includes the 674 human IG and TR genes (1245 alleles). Five hundred ninety-six genes (1139 alleles) are organized in 7 loci on 4 chromosomes, spanning a total of 6 megabases: IGH (14q32.33), IGK (2p11.2), IGL (22q11.2), TRA (14q11.2), TRD (14q11.2), TRB (7q34) and TRG (7p14) (The Immunoglobulin FactsBook, 2001; The T cell receptor FactsBook, 2001). Seventy-eight orphans (106 alleles) are found outside the main loci. IMGT® gene data are provided on Ensembl Genome Browser (EBI) via a DAS server. IMGT/GENE-DB gene entries are cross-referenced by HGNC database, GenAtlas, Entrez Gene (NCBI) and Vega (Wellcome Trust Sanger Institute). IMGT/GENE-DB reference sequences are crucial for the assignment of new alleles of IG and TR from different haplotypes (1,000 genomes project), for gene expression studies in normal and pathologic situations (cDNA high-throughput sequencing) and for biotechnology related to antibody engineering and antibody humanization.