

# ***Analysis of HLA Class I non-coding regions***

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# Introduction

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- Sequencing of HLA non-coding regions has been vastly overlooked.
- There has been an assumption that there is minimal functional significance (except at splice sites).
- Until recently, analysis of non-coding regions has been technically challenging.
- In a recent unpublished study several novel alleles within non-coding regions of HLA class I (with polymorphisms in non-coding regions) were reported.
  - Eg: Three “new” differences in the non-coding regions of HLA-B\*18:01:01 were identified in intron 3, 5, and the 3' UTR. The intron 3 region was sequenced in 48 samples with B\*18:01:01. It was found that the novel polymorphism was present on B\*18:01:01 of the 18.1 Ancestral Haplotype (A\*25, B\*18, DRB1\*15:01). The alternative polymorphism was found on the 18.2 AH (A\*30, B\*18, C\*05, DRB1\*03:01).
- Whilst some polymorphisms may not have a direct functional significance, they may be markers for specific ancestral haplotypes. These may be utilised in donor selection for unrelated bone marrow transplantation.
- At the very least we will be able to determine the level of heterogeneity in population samples.

# Proposal

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- The aim is to identify novel HLA class I non-coding variations in different populations.
- Participants will analyse SBT results for non-coding polymorphisms.
- A summary of the numbers of loci, alleles and non-coding sequences analysed to be tabulated.
- Catalogue the number of novel alleles including the HLA types at other loci for haplotype characterisation.
- Submit data for collation.
- Present data at the 16th IHIW.

## **Laboratory Requirements**

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- Ability to perform SBT and/or access to HLA Class I sequence data.
- Software capable of complete gene analysis.

## **Time Line**

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- Last enrolments late October 2011
- Distribute participant survey mid Nov 2011
- Return surveys by early Jan 2012
- Collate data by mid April 2012
- Present final data at 16IHIW