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# Frequency & heterozygosity assessment of STR markers linked to β-globin gene applicable in PND &PGD

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## Introduction

β-Thalassemia is one of the most common hereditary hemoglobinopathy disorders with a high frequency in Iran .For couples with high risk of transmitting a genetic defect, Pre-implantation Genetic Diagnosis (PGD) can be an alternative way to the select healthy embryo/s before implantation in the uterus. Homozygosity mapping with the help of STR markers in conjunction with direct sequencing can be a very powerful technique to select the best embryo. Using highly polymorphic marker is very helpful in detecting the very important event of "Allele Drop out" in PGD procedure. This study aimed to find polymorphic STR markers linked to the b-globin gene for use in homozygosity mapping studies in of PND (Prenatal Diagnosis) & PGD studies.

## Methods

50 unrelated individuals were genotyped to assess the allele frequencies, heterozygosity of the selected markers. Polymorphic STR markers were selected from Tandem Repeats Finder and Sequence-based Estimation of Repeat Variability databases.

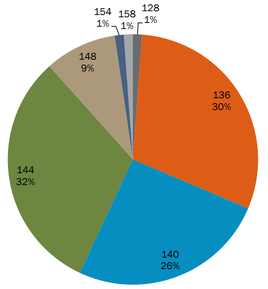
Suitable primers were designed to be to set up a multiplex-PCR reaction. Genotyping of each individual were performed using fragment analysis by ABI Genetic Analyzer 3130.

Statistical analysis was performed using GenAlEx6.03 softwere.

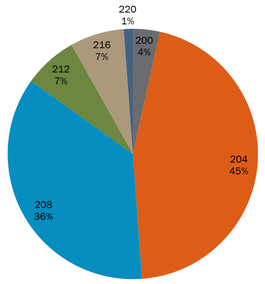
## Results

Our results showed that the heterozygosity of selected markers were between 67%-80%. Totaly, 43 alleles were observed. The highest heterozygosity was observed for D11HBBSU6.1 and the lowest for D11HBBSU15.6.7 ,6,7,5,11,6 different allele were seen for D11HBBSD3.3, D11HBBSU2.9,D11HBBSU6.1,D11HBBS D11.2, D11HBBSU11, D11HBBSU15.6 respectively.

Allele Frequency at D11HBBSU6.1 for Pop1 (n=50)



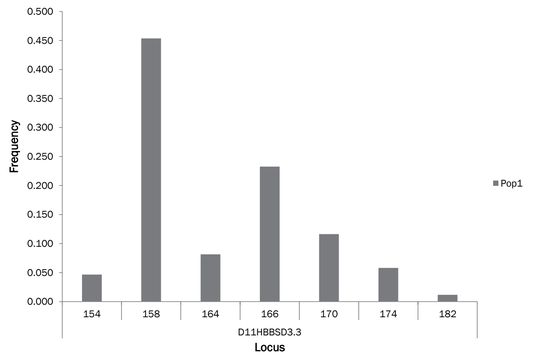

Allele Frequency at D11HBBSU15.6 for Pop1 (n=50)




## Conclusion

All studied loci were in Hardy-Weinberg equilibrium except for D11HBBSU11 and D11HBBSD3.3. The deviation could be because of high number of alleles in these loci.

Allele Frequency for D11HBBSD3.3




Allele Frequency for D11HBBSU11