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Abstract Topic:- Molecular and cytogenetic diagnostics

Abstract Title:- Novel High Throughput KASP genotyping for Sickle Cell Anaemia in Gujarat and Madhya Pradesh Tribes

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Aims:-To develop a novel Kompetitive Allele-Specific PCR (KASP) assay for the accurate and value-powerful genotyping of sickle cell mutation.

Methods:- The present study involves the screening of Sickle cell Allele in tribal population of Gujarat and Madhya Pradesh using KASP based approach. The genotyping results were further compared with PCR-RFLP and validated using Sanger sequencing.

Results:- Our findings indicate a 100% concordance between KASP-based genotyping and sequencing results. Additionally, we observed 100% concordance between PCR-RFLP and KASP. We observed a prevalence of 16.41% SCT (HBAS) in Dongri Bhil, 4.67% in Kol, and 9.33% in Korku, with a 0.7% prevalence of SCD (HBSS) in Dongri Bhil.

Conclusions:- This study first time reports the development of a novel cheap, robust and efficient Kompetitive allele-specific PCR (KASP) assay for sickle cell genotyping. The early, precise, and cost-effective molecular screening of sickle gene mutations in various tribal populations in India offers the potential to facilitate early identification, effective management, and the eventual elimination of this debilitating disease.

Keywords:- Kompetitive allele-specific PCR (KASP), Sickle cell disease (SCD), Sickle cell Trait (SCT), tribal population