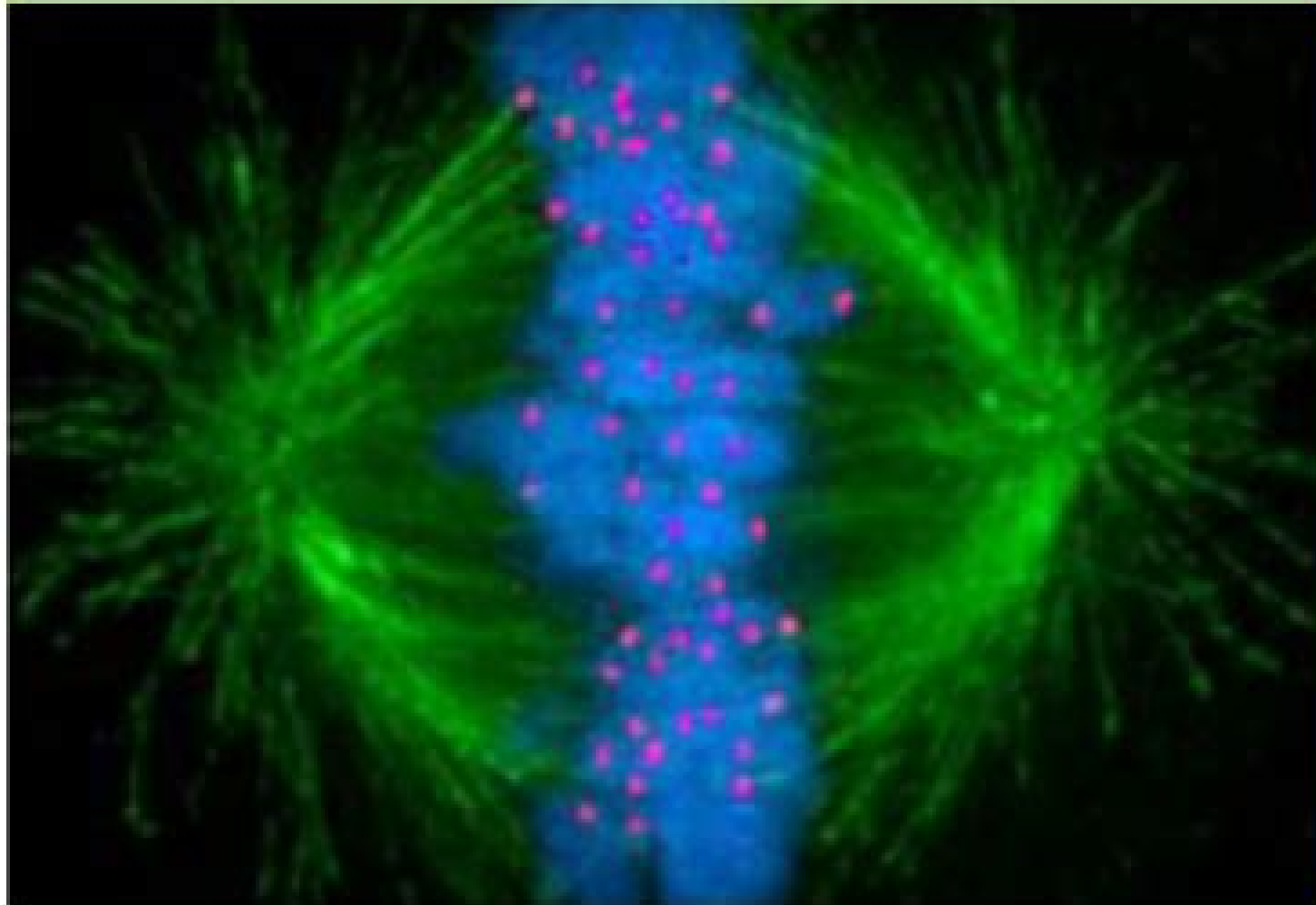


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Mapping and DNA sequence analysis of Genes underlying Isolated Autosomal Human Hereditary Alopecia in Pakistani families Khushbakht Khan, Sehrish Khan, 2014-12-23 Bachelor Thesis from the year 2014 in the subject Biology Micro and Molecular Biology grade 1 International Islamic University language English

abstract Alopecia is a broad term including many forms of hereditary hair loss resulting from genetic defects affecting hair growth cycle or hair structure that vary in age of onset severity and associated ectodermal abnormalities The inheritance pattern of alopecia can be autosomal dominant autosomal recessive or X linked Various mutations in several genes on different chromosomes are being identified which are involved in pathogenesis of inherited autosomal recessive alopecia In present research two families A B with isolated hereditary alopecia residing in different zones of Pakistan were ascertained The mode of inheritance inferred as autosomal recessive One family was subjected to mutation screening while on other polymorphic microsatellite markers was used for the purpose of homozygosity mapping to explicate the gene defect Phenotypic analysis of family A shows the characteristic clinical features of hypotrichosis with sparse hair on head and rest of body and with no associated abnormality Gene linked to this family in previous research was CDH3 So splice junction site and sixteen exons of this gene were sequenced but were negative for functional sequence variant This clearly shows mutation must be present in regulatory region of this gene In family B affected individual s shows clinical features of atrichia with papular lesions APL which is rare autosomal recessive disorder characterized by occurrence of complete hair loss with the development of keratin filled cysts Known candidate genes DSG4 HR LIPH and LPAR6 were tested for homozygosity mapping via polymorphic microsatellite markers Genotyping data showed no linkage to any of the candidate loci and therefore their involvement in causing atrichia with papular lesions in this family is not supported

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