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# Investigation of Hereditary Primary Microcephaly in Consanguineous Families

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Genetic studies mainly focus on identifying mutations at nucleotide level, causative genes for that particular disorder which are basically the output of consanguineous marriages in most of the cases especially in Asian population. For the present study genetic disorder i.e. *Hereditary Primary Microcephaly in Consanguineous Families* has been selected to investigate the linkage analysis and variant mapping. Hereditary primary microcephaly (MCPH) is a neurological disorder in which prenatal brain growth is significantly reduced. Moreover, MCPH is identified by measuring circumference of head from the forehead to the occipital prominence at the back of the head. MCPH is genetically heterogeneous mapped to different regions (MCPH1-MCPH10) including *NDE1* gene. MCPH encoding genes are involved in regulating cell cycle checkpoint, in DNA repairing events, centrosome related functions, spindle formation, kinetochore attachment to spindle and apoptosis. Its incidence rate is found to be high in consanguineous population. Abnormal spindle-like microcephaly associated (*ASPM*) gene accounts for most of the cases of MCPH. About more than 85 mutations have been reported in *ASPM* so far, including non-sense, frame shift and splice site mutations. Present study involves genetic mapping in three consanguineous Pakistani families (A, B, C) with hereditary microcephaly. These families were characterized by typical features of MCPH, having reduced head circumference along with sloping forehead in most cases. Linkage in these families was tested by microsatellite markers for the causative MCPH genes. Family A was excluded from all the respective MCPH loci where as Family B, C depicts linkage to the MCPH1 and MCPH5 locus. Sequence analysis of coding exons of *Microcephalin,* at MCPH1 was carried out in an affected member (V-5) of family B but failed to identify any functional sequence change in the coding region of this gene signifies the involvement of regulatory region. In family C, coding exons (15, 16, 17) of *ASPM* gene are selected for exon sequencing in an affected member (IV-2). DNA sequence analysis revealed a G to A transition at nucleotide position 3978, producing immediate premature stop codon (p.Trp1326*) in exon 17 of the *ASPM* gene. In carrier member (III-1) sequencing result revealed heterozygosity for this sequence variant.

## Biography

Fouzia Kausar has completed her Mphil from Quaid-i-Azam University, Pakistan in Biocehmistry. Human Molecular Biologist with 4 years national, international research experience in molecular genetics. Currently, she is working as Lecturer in the department of Biotechniology, Women University of AJ&K, Pakistan.