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LINKAGE ANALYSIS OF AUTOSOMAL RECESSIVE PRIMARY MICROCEPHALY (MCPH) IN PAKISTANI FAMILIES

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The work in this study is done on the inherited disease autosomal recessive primary microcephaly (MCPH). MCPH is characterized by reduction in head circumference (3-4 standard deviations below age and sex average) caused by underdevelopment of the fetal cerebral cortex. This is accompanied with variable degrees of mental retardation. MCPH demonstrate genetic heterogeneity and to date eight loci (MCPH1-MCPH8) and eight genes have been identified. These MCPH genes are involved in important cellular functions mainly related to mitosis of neurons (neurogenesis); cell cycle regulation, organization and orientation of mitotic spindle fibers and centrosomal production of microtubules. In the present work, two families showing primary microcephaly were studied. DNA samples from affected and normal individuals from both families were tested for linkage to known MCPH loci. All known loci were excluded from both families because of absence of linkage and homozygosity. The disease loci on some other cytogenetic location are yet to be discovered.

Biography

Iram Naz has completed her MBBS in 2014 from Xi'an Jiaotong University, China and now is a Master's (Pediatrics) student in Xi'an Jiaotong University.

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