World Academy of Science, Engineering and Technology
International Journal of Bioengineering and Life Sciences
Vol:11, No:08, 2017

# Screening of the Genes FOLH1 and MTHFR among the Mothers of Congenital Neural Tube Defected Babies in West Bengal, India

**Authors :** Silpita Paul, Susanta Sadhukhan, Biswanath Maity, Madhusudan Das

**Abstract :** Neural tube defects (NTDs) are one of the most common forms of birth defect and affect ~300,000 new born worldwide each year. The prevalence is higher in Northern India (11 per 1000 birth) compare to southern India (5 per 1000 birth). NTDs are one of the common birth defects related with low blood folate and Hcy concentration. Though the mechanism is still unknown, but it is now established that, NTDs in human are polygenic in nature and follow the heterogeneous trait. In spite of its heterogeneity, polymorphism in few genes affects significantly the trait of NTDs. Polymorphisms in the genes FOLH1 and MTHFR plays important role in NTDs. In this study, the polymorphisms of these genes were screened by bi-directional sequencing from 30 mothers with NTD babies as case. The result revealed that 26.67% patients had bi-allelic FOLH1 polymorphism. The polymorphism has been identified as p.Y60H and frequent to cause NTDs. The study of MTHFR gene showed 2 different SNPs rs1801131 (at exon 4) and rs1801131 (at exon 7). The study showed 6.67% patients of both mono- and bi-allelic MTHFR-rs1801131 polymorphism and 6.67% patients of bi-allelic MTHFR-rs1801131 polymorphism. These polymorphisms has been responsible for p.A222V and p.E429A change respectively and frequently involved in NTD formation. Those polymorphisms affect mainly the absorption of dietary folate from intestine and the formation of 5-methylenetetrahydrofolate (5 MTHF) from 5,10-methylenetetrahydrofolate (5,10- MTHF), which is the functional folate form in our system. Though the study is not complete yet, but these polymorphisms play crucial roles in the formation of NTDs in other world population. Based on the result till date, it can be concluded that they also play significant role in our population too as in control samples we have not found any changes.

**Keywords :** neural tube defects, polymorphism, FOLH1, MTHFR
**Conference Title :** ICLBS 2017 : International Conference on Life and Biomedical Sciences
**Conference Location :** Amsterdam, Netherlands
**Conference Dates :** August 07-08, 2017