32nd World Pediatrics Conference

December 04-05, 2019 | Barcelona, Spain

Study of certain genetic polymorphisms of folate and homocysteine metabolism and neural tube defects

Abbas Amel
University of Ouargla, Algeria

Neural tube defects (NTDs) represent a major cause of infant mortality among congenital malformations whose pathogenesis remains poorly understood. In this study realized on an Algerian population, we investigated the frequency of many polymorphisms involved in the of folate/homocysteine metabolism, their possible contribution in the etiology of NTDs, as well as their influence on folate and homocysteine concentrations.

The study involved children with NTD, mothers who had a child with NTD and a control population. The mutations were determined by the PCR/RFLP method while the assay of the biochemical parameters was performed by chemiluminescence.

The analysis of genetic polymorphisms has shown that C677T polymorphism in MTHFR gene affects homocysteine metabolism in mothers of NTD cases leading to homocysteine concentration values higher in mothers with TT genotype of the C677T ($p < 0.05$).

For the polymorphism A2756G of the MTR gene, although, its association with NTD risk appears to be negative, It was found to decrease the RBC folate level strengthen the contribution of A2756G mutation in NTDs occurrence ($p <0.05$).

The major challenge remains translating study observations into a clinical setting such as gene screening in women before pregnancy, to identify predisposition and seek targeted prevention. Measurement of homocysteine concentration may also be a useful indicator of the need for pre-conception intervention. There is optimism that food is an element on which intervention possibilities exist.