Analysis of Causative Factor Contributing to Recurrent Neural Tube Defects: A Case Report

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Neural tube defects (NTD) become a concern in developing countries, with rare recurrence risk 2-5%. Folate and cyanocobalamin deficiencies are the common cause of NTD, while other cause are MTHFR enzyme mutation and the presence of Maternal Serum Folate Reseptor Auto-antibodies. Case Description: A multigravida (GIV P3001), 35 year old mother having anencephalic babies on second and fourth pregnancy with was examined at antenatal clinic Dr. Soetomo Hospital. She was tested negative for TORCH, Ultrasound examination revealed positive Frog sign, cerebellum located outside cranial lobe with cervical spine defect to upper femoral region. Termination of pregnancy was performed, a female 300 gram stillbirth baby was delivered. Open upper cranium was observed with intact brain structure and non closure of cervical spine. After postpartum period, maternal serum folate and cobalamin level were observed within normal values. Maternal serum homocysteine levels was also observed. MTHFR enzyme has a critical role at converting methylentetrahydrofolate to active tetrahydrofolate, and converting homocysteine serum to methionone used for protein synthesis. The mutant MTHFR C677T have 30% lesser converting function in metilathion chain. The decrease of this conversion in metilathion proccess resulting in increase unconverted homocysteine serum level accumulation. The serum of maternal NTD's pregnancy contain Folate Reseptor Auto-antibodies that binds to folate receptor and blocking cellular folate uptake. Conclusion: Mother with history of NTD pregnancy should encourage to take routine daily 400-800 mcg folic acid supplementation, routine antental care is mandatory, ealy ultrasound examination should be performed for monitoring and diagnosis.

Biography: Margaretha Claudhya Febryanna, has completed her MD at the age of 24 years from Airlangga University, then worked for four years in Bojonegoro, East Java as primary doctor in Public Health Centre and now continue her study as the resident of Obstetric and Gynecology in Airlangga University, Dr. Soetomo Hospital, a tertiary hospital in Surabaya, Indonesia.