

Correspondence

Dr Gehanath Baral Kathmandu University, Nobel Medical College Teaching Hospital, Biratnagar, Nepal

Email:

baraldr@gmail.com Phone: +977-9841228829

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Neural tube defects: An Obstetrician's concern

Gehanath Baral

Kathmandu University, Nobel Medical College Teaching Hospital

ABSTRACT

Screening for neural tube defect is a universal procedure that doesn't require any criteria; but the intervention method is altered. Folic acid intervention is the proven method and a good ultrasound is the simple equipment to screen.

Keywords: birth defects, folic acid, screening, ultrasound

COMMENTS

Birth defect of brain and spinal cord is called the neural tube defect (NTD) and 70% of it can be prevented with a simple proven intervention like Folic acid (Vitamin B9) supplementation. This defect is the event of first month of pregnancy; thus, the intervention would be missed prior to knowing the pregnancy. NTD screening is the universal procedure since it occurs in 90-95% cases without known risk factors.¹

Causes of NTDs are genetic, environmental and autoimmune factors by disrupting the neural tube closure pathways.^{2,3} Malabsorption syndrome, nutritional deficiency, hemolytic anemia, antifolate medicines (like antiepileptics and methotrexate) and Diabetes mellitus may cause NTDs.^{4,5} Febrile illness during pregnancy and obesity may also result in miscarriage and birth defect especially NTD, cardiac and lip defects.^{6,7}

Supplementation of 400mcg daily from one month prior to pregnancy through 12 weeks; and from 3 months before pregnancy with 10 times more dose if high risk (history of NTD in either partner) for NTD.^{8,9} Folic acid, alone or in combination with vitamins and minerals, prevents NTDs, but does not have a clear effect on other birth defects.¹⁰

The red blood cell folate threshold can be used as a population level indicator of folate insufficiency in women of reproductive age and should be above 400 ng/mL (906 nmol/L). Folate concentration <3 ng/mL in serum and <100 ng/mL in RBC are likely to be indicator of folate deficiency resulting in megaloblastic anemia. There is less biological variation in RBC folate level. To prevent NTDs where the pregnancy occurs unnoticed, the multivitamin B combined with choline, betaine and *n*-3 PUFAs supplementation may have a better protective effect against NTDs than folic acid alone. 12

High resolution

ultrasonography alone at 11-14 weeks (TVS) and 18-20 weeks (TAS) would be better than MSAFP to detect NTDS and trisomy 18 and 21 as well. MSAFP assay is less sensitive in first trimester; so, it can be performed from 15 weeks through 20 weeks. MSAFP assay is less sensitive in first trimester.

CONCLUSIONS

Screening of neural tube defect should be a routine practice for the obstetrician and the Folate supplementation is the must to start prior to one month of pregnancy to prevent it. A good resolution ultrasound would be better than the biochemical markers as the screening tools.

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