Folic Acid Deficiency and Neural Tube Defects: Catastrophic Consequences

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Abstract

Neural tube defects are conditions of malformation in the embryonic formation of the spine. They are very distressing and cause preventable deaths and disability. Folate is a key ingredient in rapidly developing cells. Folate deficiency causes a decrease in serum concentration, erythrocyte folate concentration and this affects rapidly dividing cells causing neural tube defects. The consequences of folate deficiency can be catastrophic causing terminations of pregnancy, neonatal deaths or permanent disability. Folic acid alone or in combination with other vitamins and minerals can reduce the prevalence of neural tube defects. Food fortification may help reduce unnecessary loss of life.

Keywords
Folate deficiency; Neural tube defects; Avoidable deaths; Permanent disability; Food fortification

Introduction

Neural tube defects are congenital malformations of the spine during its embryonic formation. They cause loss of lives, permanent disability and psychological trauma to the parents. They are associated with folic acid deficiency, and therefore may be preventable. Patients and clinicians need to be made aware of the importance of taking preventative folic acid periconceptionally. This may be difficult as half the pregnancies are unplanned.

Folate is a key ingredient in rapidly developing cells. It is needed in cell growth, cell division, cell synthesis and repair of DNA. During pregnancy fetal growth causes an increase in the number of rapidly dividing cells which leads to increased folate requirements. Folate is a water soluble vitamin found in certain fruits and foods. A diet low in fresh fruits and vegetables is the main cause of folic acid deficiency. Folic deficiency is associated with neural tube defects such as spina bifida. Affected infants may suffer life-long disabilities. They may be paralysed from the below the spinal defect.

Discussion

Folate deficiency causes a macrocytic anaemia with megaloblastic changes in the bone marrow. There is a raised mean corpuscular volume of more than 80 fentolitres on a full blood count. The diagnosis is confirmed with reduced folate levels in the serum and red cells.

Consequences of lack of folate include orofacial clefts, gastrochisis and neural tube defects [1]. Neural tube defects include anencephaly, spina bifida and encephalocele [2]. Neural tube defects are preventable causes of morbidity and mortality [3]. Prenatal diagnosis is mainly by detailed ultrasonography at 18-20 weeks gestation. All pregnant women should ideally undergo a detailed anatomy ultrasound scan at 18-20 weeks gestation to rule out such congenital malformations. In affected fetuses there is a 90% chance of the pregnancy being terminated [4]. This causes deep distress and unnecessary loss of lives.

Periconceptional folic acid significantly reduces the risk of neural tube defects [5, 6]. In high income countries where there is folic acid supplementation in the periconceptional period and early pregnancy, there has been significantly reduced adverse outcomes like birth defects [7, 8] Not all the neural tube defects may be amenable to folic acid supplementation as the cause may be multifactorial including environmental effects [9].

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In women planning a pregnancy an intake of 0.4mg of folic acid until at least 13 weeks after conception may prevent neural tube defects [10]. Those at increased risks of neural tube defects like patients on anti-epileptic drugs or with previously affected babies, a dose of 4mg is recommended. Public awareness via information dissemination [4] and use of pharmacists [10] can help inform women in person about the benefits of folic acid supplementation.

Mandatory food fortification in flour with folic acid in Brazil has helped significantly reduce the prevalence of neural tube defects [11]. Countries that have not introduced food fortification like the U.K, still have avoidable terminations of pregnancy, stillbirths, neonatal deaths and permanent serious disability in surviving children [12]. Surviving children need specialised care and expert surgery [13].

**Conclusion**

Folic acid alone or in combination with other vitamins and minerals reduces the prevalence of neural tube defects [14]. Mandatory food fortification worldwide may help reduce the burden of avoidable deaths and disability.

**Competing Interests**

None

**References**


