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Arnold chiari malformation with spina bifida

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Abstract

In Arnold chiari (kee-AHR-ee) II malformation elongated cerebellar tonsils are displaced inferiorly through the foramen magnum in to the upper cervical spinal canal. It is a complex anomaly with skull, dura, brain, spine and cord manifestations. Meningomyelocele is seen in all cases. We present a case of type II Arnold chiari malformation diagnosed in utero at 2nd trimester. There was no periconceptional folic acid supplementation. As the role of methylene tetra hydro folate reductase gene polymorphism in neural tube defects is becoming evident, a simple opportunity as folic acid supplementation should not be missed. Folate supplementation as fortification of cereal grains will also prevent other conditions like congenital heart defects, urinary tract anomalies, orofacial defects, limb defects and pyloric stenosis.

Keywords : Arnold chiari, Folic acid, Pregnancy, Periconception

Case report

A 29 yr old lady presented to the antenatal clinic at 21 week gestation for routine antenatal check up. Her last menstrual period was 6 months back. She was married for 8 yrs. She was a primi gravida with infertility treated (ovulation induction 5 cycles 2 yrs back). Her cycles were irregular. This was a spontaneous conception and pregnancy was confirmed after 14 weeks by USG. She was moderately pale and had not taken any folic acid or iron supplementation. On examination symphysio-fundal height corresponded to 20 week of gestation. Fetal movements were present and fetal heart was heard. Her haemoglobin was 9gm/dl.

Obstetrical ultrasonography of fetal cranium revealed a crowded posterior fossa and dilated supratentorial vascular system suggestive of hydrocephalous (Fig - 1). The spinal canal in the cervical region was dilated. There was absence of spinous process in the lumbar spine. There was open spinal dysraphism with dorso lumbosacral myeloschiasis. All these findings suggested type II Arnold chiari malformation. Consent for medical termination of pregnancy was taken after counseling the couple. MTP was undertaken with vaginal misoprostal 2 tablets. A male foetus of 700 grams was expelled vaginally. Cord blood was collected for karyotyping. The foetus had hydrocephalous with a swelling in the posterior occipital region. Open spinal cord with meningomyelocele was present in lumbosacral region (Fig-2). Patient was discharged on folate and iron supplements. Fetal karyotype study was normal.

Discussion

Arnold Chiari malformations were first described in pediatric autopsy specimens in 1891 by Hans Chiari -Austrian Pathologist (1851-1916) 1. In legacy with the name of his professor Dr Arnold, and his name, the hind brain disorder is named as Arnold Chiari malformation 2. The incidence is gradually rising because of increased detection with MRI. Exact etiology so far is unknown. Chiari I malformation, the tonsillar ectopia is mostly asymptomatic until adulthood. The diagnosis is made by measuring how far the tonsils protrude below the margin of foramen magnum. The measurement is taken from (ophisthion to basion) inner margins of foramen magnum to the inferior most part of tonsils. A simple rule 3 is

- Above the foramen magnum : normal
- < 5mm : also normal but the term benign tonsillar ecto-pia can be used
- >5 mm : Chiari I malformation

It should also be noted that the “normal’ position of cerebellar tonsils varies with age 4. In neonates the tonsils are located just below the foramen magnum and descend further during childhood, reaching their lowest point somewhere between 5-15 years of age. As the age advances the tonsils ascend and come to rest at the level of foramen magnum 4.

In Chari II malformation, the pegs like cerebellar tonsils are displaced inferiorly through the foramen magnum into the upper cervical spinal canal. The vermis the fourth ventricle and medulla are normal or minimally deformed. Moderate hydrocephalous has been documented in about 25% cases 2. Syringomyelia is present in 60% of established cases. Skeletal anomalies are found in 25% of cases. Basilar invagination are found in 50% of cases, Klippel Feil syndrome is present in upto 10% of cases. Atlanto-occipital assimilation can also be present.

Skull malformations may include calvarial defects (lacunar skull). There is small posterior fossa with low lying transverse sinuses. The falx is fenestrated and the tentorium hypoplastic. Foramen magnum is wide. Brain anomalies include inferiorly displaced vermis, medullary kink and spur. Corpus callosal hypoplasia or complete agenesis may be present. Myelomeningocele is present in all cases. We speculate that hind brain herniation is secondary to meingomyelocele. This results as a consequence of failed posterior neural pole closure. The posterior neural pole usually closes at 6 wk of menstrual age 5.

The Chiari III malformation includes herniation of hindbrain into the low occipital and high cervical region along with Chiari II malformation 2. In rare cases the pons and medulla may also be herniated. The herniated brain tissue may show necrosis, fibrosis and gliosis. The Chiari IV malformation is a case of severe cerebellar hypoplasia. The posterior fossa is large, the brain stem is small and there is no hydrocephalous 2.

In view of the severe morbidity associated with these malformations, a daily intake of 400µgms of folic acid is recommended in the preconception period. The Methylene tetrahydrofolate dehydrogenase gene (MTHFD is one of the key genes involved in folate pathway 6. A large metanalysis study strongly suggests that the MTHFD 1 G1958A gene polymorphism is strongly related to neural tube defects 6. Genetic defects are not modifiable. Albeit, a lot can be done to fortify folic acid supplementation in pregnancy to reduce the incidence of neural tube defects. Folic acid is heat labile. Avoiding overcooking, including fresh green leafy vegetables in diet and cereal grain fortification of folate can reduce the incidence of all folate deficiency associated congenital anomalies.

We decided to terminate the pregnancy after prognosticating the parents. Although a preconceptional folic acid supplementation could have been done in our set up, this was missed, as the lady presented late in the pregnancy. Majority of neural tube defects can be prevented by folic acid supplementation in the preconceptional period. Folic acid fortification of staple food grains has lead to reduction of incidence of neural tube defects in USA, Canada, Mexico and Nova Scotia 8-11.

In Europe, the European Registration of Congenital Anomalies and Twins (EUROCAT) has published a special report on the prevention of neural tube defects by periconceptional folic acid supplementation 12. Associations of Arnold Chiari malformations with low serum Vitamin B12 levels have been documented in two studies 13,14.

It is believed that 1; 00,000 years ago our ancestors ate 14-16 servings of fresh fruits and vegetables (major source of folic acid) each day 5. With our modern cooking, we are all at a risk of inadequate folic acid intake through diet alone. Folate is required to synthesize RNA and DNA, required for growth, repair and adequate immune response. We should have adequate folate as children for appropriate growth, as women to prevent birth defects and as adults for proper cardiovascular health and cancer protection. The best possible strategy would be fortification of staple cereal grains with daily intake of 400µg folate. A potential effect would be unmasking of background Vitamin B12 deficiency. This will need attention especially in elderly population to prevent neuropathy. A dual fortification with folate and Vitamin B12 can be considered in India as well. The opportunity is open.

Regular folic acid undergoes a 4 – step enzymatic conversion process to become the biologically active L-methyl folate – the form of folic acid used by the body.

(FOLIC ACID à DIHYDROFOLATE à TETRAHYDROFOLATE à METHYLENE TETRAHYDROFOLATE à L - METTHYL FOLATE)5.

The final step of conversion involves MTHFR enzyme. Unfortunately , over 50% of people have MTHFR enzyme dysfunction leading to inappropriate conversion of folic acid to LMF which leads to significant reduction in the amount of L- methyl folate availability to the body. L-methyl folate – the active form of folic acid which can be immediately absorbed by the body and does not depend upon the enzyme MTHFR to get converted to its active form. Therefore, more than folic acid, administration of L-methyl folate helps to attain the full benefit of supplementation, even with those who have deficiency of MTHFR5.

Conclusion

Arnold Chiari Malformations can be prevented by preconceptional folic acid (more preferably L-methyl folate) and Vitamin B 12 supplementation. In India, most of the pregnancies are unplanned. The rostral and caudal neural pores close at 6 wk of gestation. A delayed folic acid supplementation is bound to miss the vital period of organogenesis and neural tube closure. It is imperative to supplement our cereal grains with folic acid and vitamin B12 to prevent the social and economic burden of birth defects.

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