RECURRENT CONGENITAL ANOPHTHALMIA- PREVENTION BY PERICONCEPTUAL VITAMIN-A

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Introduction

A series of studies made in the past few years have provided fresh perspective on the role of vitamin A in prevention of congenital anomalies [1,2]. We are reporting a case of recurrent anophthalmia, with a favourable outcome after administration of periconceptual vitamin A, as an evidence to support this concept. Vitamin A has a number of important functions in the body. It is necessary for growth and differentiation of epithelial tissues, growth of bone and normal functioning of the retina. One of the more important functions of Vitamin A is its role in embryonic development. Vitamin A deficiency during pregnancy may be associated with human teratogenesis like microcephaly, anophthalmia, microphthalmia and coloboma [3].

Case Report

A twenty-six-year old lady delivered a malformed baby in December 1992. The baby had multiple congenital anomalies, such as bilateral absence of eyeballs (anophthalmia), cleft lip and palate and congenital heart disease. The baby expired after 48 hours. The patient was investigated fully. There was no history of consanguineous marriage, drug intake (apart from iron, folic acid and calcium) or any radiation exposure during pregnancy. There was no positive family history of congenital abnormalities in the previous two generations. Routine blood and urine tests, blood sugar levels, TORCH titres, VDRL, placental histo-pathological examination and karyotyping done both for husband and wife, were found to be normal. Since no cause was found, the patient was advised to take tablet folic acid 5 mg daily, from three months prior to conception, empirically. The patient conceived a second time, 3 months after folic acid. This pregnancy also progressed normally, but the patient again delivered a malformed baby, a male weighing 2.7 kg in May 94. The congenital abnormalities were similar to those in the first baby. This baby also expired after 48 hours. All the investigations which were done after delivery of the first baby were repeated, but were again found to be within normal limits. This time the patient was asked to take vitamin A 5000 units along with folic acid supplements, daily, before conception. The patient conceived after 4 months of this treatment. Vitamin A supplements along with folic acid and iron were continued throughout this pregnancy. The patient delivered normally, at 39 weeks of gestation, a male child weighing 2.8 kg in January '96. This baby didn't have any

congenital anomalies.

The patient conceived again in 1998, this time without periconceptual vitamin A supplement. She came for her first antenatal check-up at 16 weeks of pregnancy. She was monitored with ultrasound (USG) for specific congenital abnormalities. USG could confirm multiple abnormalities in the foetus (anophthalmia, cleft lip and palate) at 24 weeks of pregnancy. In August '98 the pregnancy was terminated. A stillborn male baby weighing 700 g was delivered. The abnormalities detected by USG were present in the baby.

Discussion

Anophthalmia is a rare congenital abnormality. Bermejo et al [4], after studying over one million births, reported its occurrence at the rate of 21.34/100,000 births. Recurrent anophthalmia is rarer. The cause of this defect could be a chromosomal defect, a single gene disorder or an environmental effect. In most of the patients, like in this case, no cause is found and therefore the treatment remains empirical. Preventive efficacy of periconceptual folic acid in prevention of neural tube defects is now well established; this fact prompted many workers to study the effect of periconceptual vitamin supplements in prevention of birth defects. Yang et al [5] reported significantly lowered congenital abnormalities in the babies born to the mothers who were given periconceptual multivitamin supplements. Czeizl [1] in a large study reported lower incidence of birth defects in patients who were given vitamin A during first trimester of pregnancy. However, routine use of vitamin A supplements during pregnancy has not become popular because of its possible teratogenic effect on the foetus, although many studies have proved that vitamin A is teratogenic only in doses above 10,000 unit daily [6]. In this case, during the first pregnancy, folic acid was given after pregnancy had occurred; in the second, periconceptual folic acid was given; in the fourth, no supplement was given in the first trimester; and all these pregnancies resulted in malformed babies. However, during the third pregnancy periconceptual vitamin A was given

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and the patient had a normal baby. This shows that there may be a possible role of vitamin A in the prevention of birth defects. We had planned to give periconceptual vitamin A to the patient during her future pregnancies, to prove our point more conclusively. But the patient had gone through immense mental trauma and did not want to be pregnant again.

The routine use of vitamin A during pregnancy cannot be advocated on the basis of just one case but one may use it in the prevention of recurrent birth defects.

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