Dizygotic Twins with Myelomeningocele

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Abstract. Among the neural tube defects incidence of spina bifida and myelomeningocele is less in twins compared to singletons. This article reports a case of dizygotic twins with myelomeningocele, a rare occurrence. Possible association of twining and neural tube defects and its impact on genetic counseling in such cases has been discussed.

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Key words : Neural tube defects; Twining; Counseling.

Prevalence of neural tube defects has been estimated to be 0.2-11/1000 births depending on geographical location. Among twins prevalence of spina bifida and myelomeningocele is reported to be less than singletons with concordance rate of about 5.1% for monozygotic twins. This article is reporting a pair of dizygotic twins with meningomyelocele in both – a rare occurrence. Implications of such presentation on genetic counseling and multivitamin supplementation have been discussed.

CASE REPORT

A pair of male twins were delivered by an emergency cesarean section for mid pelvic contracture to a primigravida at 40 weeks of gestation (Fig. 1). There was no history of consanguinity. Mother had no antenatal risk factors and did not take any antenatal care or undergo any ultrasound examination during pregnancy. She did not take any multivitamin supplementation before or during pregnancy. There was no history of twins or neural tube defects in the family. Placenta was diamniotic and diachorionic.

First twin was a 2 kg male baby born with a normal Apgar score. He had dysmorphic facies with low set ears, high arched palate, short neck as a small pulsatile swelling 2x2cms in the frontal region (Fig. 2). Spine had an open myelomeningocele in lower dorsal region. On examination, vitals were normal. Head circumference was 32 cm. Both anterior and posterior fontanelle were widely opened and all sutures were separated. Lower limbs had lower motor neuron paraplegia. Anal reflex was absent. There was bilateral congenital talipes-quinovarus. On investigation kidney function test was normal, blood group was A positive. Infantogram revealed absence of pedicle and corresponding ribs on left side of T6-T8 vertebrae. Ultrasound of abdomen was normal. Gross dilatation of lateral ventricles with moderate obstructive hydrocephalus was seen on ultrasonography of cranium. Contrast enhanced computed tomography revealed obstructive hydrocephalus at the level of cerebral aqueduct and frontal encephalocele. Child had respiratory distress since birth; was started on intravenous fluids and antibiotics. Baby had cardiorespiratory arrest on third day of life and expired in spite of all resuscitative efforts.

The second twin weighed 1.4 kg at birth, had normal...
Fig. 2. Dysmorphic facies of twin I with encephalocele.

apgar score, head circumference of 30cm. On examination facies were normal, gestation was term, all sutures were separate, lower limbs were paraplegic with absent deep tendon reflexes and anal reflex. Open myelomeningocele was present in the lower lumbar region. Systemic examination did not reveal any abnormality. Infantogram revealed spina bifida of T₁₁ – T₁₂ vertebrae with scoliosis towards right side. Ultrasonography of abdomen was normal. Ultrasonography of skull revealed dilated lateral ventricles and mild hydrocephalus. Blood group was AB positive. Computed tomography could not be performed on this child, child was started on intragastric feeds, subsequently he had cardiorespiratory arrest, succumbed on day 2 of life.

**DISCUSSION**

The epidemiology of neural tube defects (anencephaly, spina bifida, myelomeningocele and encephalocele) is complex and numerous studies have been conducted in an attempt to elucidate their etiology. Genetic, environmental and multifactorial causation has been suggested. Risk of recurrence has been reported to be 4-5% after first child with neural tube defects and 10% after two siblings with neural tube defects. Traditionally studies in twins have been carried out to elucidate the relative contribution of genetic and environmental factors but it is difficult to gather large number of twin pairs with a particular congenital anomaly. Increased incidence of congenital anomalies has been associated with monozygotic twins. Concordance rate of central nervous system anomalies in twins is very low. Review of literature does not reveal increased incidence of myelomeningocele or spina bifida in twins compared to singletons, though incidence of anencephaly has been reported to be more in twins. A large study of 96,000 live births with congenital malformations only 3 pairs of twins were concordant for spina bifida. There are only a few case reports of dizygotic twins with meningomyelocele and hence the importance of this case.

Review of literature has revealed studies implicating that twining and neural tube defects may have common genetic or environmental factors. It was observed that upper neural tube defects were associated with excess twining of monozygotic type or same sex dizygotic twins. Neural tube defects and twinning both arise early in human embryonic development and that one may be casually related to the other has been suggested by the fact that they occur together more often than expected by chance.

In familial association of neural tube defects with same sex dizygotic twins delayed fertilization has been implicated. Delayed fertilization results on over ripe ova which lack cohesion and hence splitting of zygote. Similarly neural tube defects can be caused by lack of cohesion of neurulating cells. An excess of dizygotic twining is implicated in mothers of probands with spina bifida and there is a increased rate of neural tube defects in siblings of dizygotic twins. This familial association between twining and neural tube defects would have practical implication in counseling i.e there might be an increased incidence of dizygotic twins on the maternal side of proband with neural tube defect and an increased risk of neural tube defects in siblings of dizygotic twins. Percentage by which the recurrence risk may be modified in these cases needs to be determined by further research.

Most of the population based studies have attempted to find out the etiology based on concordance rate in monozygotic twins. Preconceptual folic acid supplementation is known to prevent upto 70% of recurrences. A recent study evaluated red blood cell folate in 440 pairs of monozygotic twins and 331 pairs of dizygotic twins and found that 46% of variance in RBC folate was due to additive genetic factors, 16% due to additive genetic effects and 38% due to random environmental effects. They found that repeat measures of folate in same individual had correlation similar to that in pairs of monozygotic twins. Hence a search should be made for specific genes influencing red cell folate and thus identify individuals with increased risk of neural tube defects in a population. A trial carried out in 5,502 pregnant women demonstrated that periconceptional multivitamin supplementation with or without ovarian stimulation increased the rate of multiple births.