

Case Report

Recurrent hydrocephalus with neural tube defect: a case report

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ABSTRACT

The occurrence of congenital hydrocephalus in successive pregnancies is a rare event. Several modes of genetic transmission have been suggested for familial cases of hydrocephalus. The most common heritable pattern of inheritance is X-linked recessive aqueductal stenosis which carries 25% risk of recurrence in future pregnancies with 50% risk for male fetuses. In families where multiple siblings of both the sexes are affected, an autosomal recessive inheritance due to consanguinity has been an alternative explanation. We report a case of female baby with congenital hydrocephalus and lumbar myelomeningocele who was born of a mother with a history of male babies with congenital hydrocephalus in her previous two pregnancies. She had first degree consanguinity with her husband. We discuss the possible differential diagnoses and etiological factors.

Keywords: Recurrent congenital hydrocephalus, Neural tube defect, Periconceptional folic acid

INTRODUCTION

Hydrocephalus is a common major congenital malformation occurring in various populations with an incidence of 0.5-2/1000 total births.¹ There are two forms of hydrocephalus, congenital and the acquired. Congenital hydrocephalus is an etiologically diverse disorder, including environmental (infectious, teratogenic) and genetic factors as the underlying mechanism.² Familial hydrocephalus presents a very considerable morphological and etiological heterogeneity, which can be a manifestation of several autosomal dominant or recessive syndromes. The non-syndromal forms include autosomal X-linked or recessive types and these tend to be recurrent.³ We report a case of recurrent congenital hydrocephalus with lumbar myelomeningocele in a family with two previous pregnancies associated with congenital hydrocephalus in male babies. The occurrence of X-linked recessive inheritance in the first and second pregnancy and autosomal recessive inheritance in the third could be a possible explanation. The objective of this report is to highlight the need for proper counseling of such families.

CASE REPORT

A 24-year-old G₃P₂L₀ at 39 weeks 2 days of gestation presented to our casualty with complaint of pain abdomen increasing in frequency at regular intervals for the past 2 h. There was a history of previous two dead pre-term male hydrocephalic babies which she delivered vaginally at around 32 weeks of gestation within the last 3 years. She gave a history of first degree consanguineous marriage with her 30-year-old husband, 4 years ago. There was no other significant past or family history of congenital anomalies or genetic diseases. In the first trimester, she was not on regular antenatal checkup and no sonogram was done. She could not recollect whether she had taken folic acid tablet in the periconceptional period. There was no history of drug intake, fever or radiation exposure in the first trimester. On her first antenatal visit at around 21 weeks of gestation, she underwent an obstetric sonogram which revealed a single live fetus at 22 week's gestation with congenital hydrocephalus and lumbar myelomeningocele. She was counseled regarding the prognosis of the fetus, but she continued the pregnancy.

At the time of admission, general and systemic examination revealed no abnormality. She was in late first stage of labor with cervix fully effaced and os 7-8 cm dilated with tense bulging of bag of membranes. She delivered a 2.9 kg live term female baby vaginally. There was poor cry of the baby following birth and the Apgar score was 6 at 1' and 7 at 5 min.

Baby was born with multiple congenital anomalies including hydrocephalus (Figure 1), lumbar myelomeningocele (Figure 2), paraplegia with bilateral congenital talipes equinovarus (Figure 3). The baby was evaluated by



Figure 1: Hydrocephalus baby.



Figure 2: Lumbar myelomeningocele.



Figure 3: Bilateral congenital talipes equinovarus with paraplegia.

paediatrician and CT scan of the brain was advised. CT scan of the brain revealed features suggestive of hydrocephalus with colpocephaly and bilateral pointed lateral horns. The rest of cerebral hemispheres and posterior fossa structures were found to be normal. Magnetic resonance imaging on day 5 confirmed the above findings of hydrocephalus. The baby was referred to Pediatric Surgery Department of the higher center for ventriculo-peritoneal shunt and myelomeningocele repair.

DISCUSSION

Hydrocephalus is one of the most common central nerves system development disorders. If left untreated, the condition leads to various degrees of cognitive impairment, cerebral palsy and visual defects. In severe cases, the condition is fatal. In developed countries, most patients undergo surgical treatment but the long term prognosis is highly variable.⁴ Neural tube defects are a variety of malformations resulting from incomplete closure of neural tube in early development of fetus and have a multifactorial etiology.

The population based study by Munch et al. found strong evidence of familial aggregation of primary congenital hydrocephalus, which supports the existence of genetic component to the etiology and the pattern of association suggests that a strong maternal component contributes to the familial aggregation.⁴ According to Zhang et al., it is estimated that X linked hydrocephalus may account for 5-15% of congenital hydrocephalus cases.⁵ The affected gene (*LICAM*), encodes L1 protein, which is a neuronal cell adhesion molecule essential for nervous system development and function. The phenotype characteristics also include adducted thumbs, agenesis or hypoplasia of corpus callosum and corticospinal tracts, mental retardation and spastic paraplegia.⁴ In our case, there is a possibility of X-linked recessive form in the first and second pregnancies, while in the third pregnancy; autosomal recessive inheritance could be the cause due to consanguineous marriage. Examples of other inherited syndromes that might contribute to the familial aggregation observed in syndromic congenital hydrocephalus include Marden-Walker syndrome (autosomal recessive) and Walker-Warburg syndrome (autosomal recessive).⁶

Viral, chemical, nutritional factors, seasonal variation on the date of conception, geographic and ethnic group differences, minor or major gene defects and consanguinity have been postulated among the etiological factors for neural tube defects which have a female preponderance. In a study by Papp et al., the recurrence risks of neural tube defects and hydrocephalus were 3.47% and 2.95%, respectively. The authors concluded that recurrence risk is mainly influenced by the pathoanatomic severity of the involved anomaly, the degree of relationship, and the number of affected relatives in the family.⁷

In fetuses with non-communicating hydrocephalus, obstruction may occur in the ventricular system in 43%

of cases. Furthermore, fetuses with communicating hydrocephalus have obstruction beyond the ventricles; 38% in the subarachnoid space and 19% in the arachnoid granulations–Dandy Walker syndrome.⁸ In cases where the obstruction is not visualized, ventriculomegaly is the most appropriate term. When the ventricles measure greater than 15 mm, many of those cases that appear isolated prenatally are ultimately found to have other abnormalities. Attention is drawn to the fact that hydrocephalus (ventriculomegaly) is commonly manifested only in the second half of gestation. Therefore, performing ultrasound examination is strongly recommended not only at the 18th but at the 24th week of gestation, as well as in pregnancies with a positive history of neural tube defects and/or hydrocephalus.⁷ However nowadays, hydrocephalus can be diagnosed very early in pregnancy on the basis of widening lateral ventricles with fetal biparietal diameter and head circumference still normal or without macrocrania.¹

Termination of pregnancy is an option for families in cases involving serious malformations that are incompatible with life. Examples of these conditions include severe chromosomal abnormalities associated with anomalies (e.g. trisomy 13), certain metabolic conditions and anatomic defects, especially of the brain and kidneys. In utero ventriculoamniotic and ventriculoperitoneal shunts for cases of isolated obstructive hydrocephalus are a few interventions available at various levels to ensure a favorable outcome.⁸ Finally, the use of folic acid 5 mg (if history of previous NTD) or 400 µg daily 3 months prior to gestation and during the first trimester of pregnancy has been reported to be beneficial in the prevention of neural tube defects.⁹

CONCLUSION

The objective of this report is to highlight the need of proper counseling of such families to prevent such recurrences in the future.

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