

# Recurrent congenital hydrocephalus: two case reports and counseling highlights

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## Summary

Familial hydrocephalus presents a very considerable morphological and etiological heterogeneity. It 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. We report for the first time in the literature on two families with recurrent congenital hydrocephalus each for the fourth time. The first showed evidence of X-linked recessive form, while the second was an autosomal recessive form. The objective of these reports is to highlight the need for proper counseling of such families.

*Key words:* Hydrocephalus; Ultrasound; Counseling.

## Introduction

Congenital hydrocephalus (CH) has been described as a heterogeneous collection of different abnormalities that result in an increase of cerebro-spinal fluid (CSF) in the cranial cavity. Its etiology can be extremely varied. It could be secondary to an open neural tube defect, a malformation of the brain, an intrauterine infection, or an intraventricular hemorrhage which may follow some trauma or tumor.

The overall incidence of CH varies from 0.2-2.0 per 1,000 deliveries [1], only one-fourth of which are said to be live births, and half of those born alive may show normal intellectual development.

Familial hydrocephalus presents a considerable morphological and etiological heterogeneity, as it can be divided into both syndromal and non syndromal entities [2]. It can be manifested as part of the phenotypes of some chromosomal aberrations such as trisomy 13, 18 and triploidy, or it can be a manifestation of several autosomal dominant diseases such as Aperts syndrome or autosomal recessive ones such as Mechel-Gruber syndrome. The non syndromal forms include X-linked or autosomal recessive form that tend to be recurrent. In general, recurrent risk for congenital hydrocephalus excluding the X-linked form is low and ranges from < 1% to 4% [1].

The autosomal recessive forms of CH have been reported in consanguineous families [3], a common practice among the Arabs, but this form has not yet been studied genetically. The reported incidence of infantile hydrocephalus has been put at 0.81 per 1,000 in Saudi Arabia, while the contribution of this to the overall incidence of childhood neuro-developmental disorders was estimated to be 4%-8% [4].

We report two rare cases with recurrent CH, each for the fourth time, which is the first time such incidence has been reported in the literature. The first showed evidence of X-linked recessive form while the second was an autosomal recessive form. The need for proper counseling for such families, needs to be highlighted.

## Case 1

A 35-year old Saudi woman, gravida 8, para 7, abortus 0, married to her second degree cousin, presented with a history of delivery of a hydrocephalic male baby who died in the early neonatal period. In addition, she had had two intrapartum fetal deaths of hydrocephalic male fetuses. She had four normal children (3 girls and one boy).

During her current pregnancy, the ultrasound at 30 weeks' gestation showed a fetus with severe isolated hydrocephalus. Induction of labor at 34 weeks of gestation ended in vaginal delivery of a live baby boy. Computed tomography (CT) revealed bilateral enlargement of the lateral ventricles, preponderant dilatation of the posterior horn, and diffuse hypoplasia of the cerebral white matter, in addition to aqueduct stenosis. No other anomalies were detected in the newborn who weighed 2.8 kg. The Apgar scores were 3 and 6 at one and five minutes, respectively, but he died 24 hours after delivery. Chromosomal analysis was normal.

## Case 2

A 34-year-old Saudi woman, gravida 11, para 9, abortus 0, married to her first degree cousin, presented at 21 weeks of gestation with an ultrasound diagnosis of isolated hydrocephalus.

She had six normal children (4 boys and 2 girls), in addition, to three babies affected by congenital hydrocephalus diagnosed antenatally; two were girls who died in the immediate neonatal period, while the third was a boy delivered by cesarean section (CS) at 36 weeks' gestation at the patient's request. The baby had a shunt operation post delivery but died after three days.

In the current pregnancy, serial ultrasound showed increasing ventricular dilatation which started at 20 weeks' gestation with

no other fetal anomalies. She was delivered at 37 weeks' gestation by emergency CS for fetal distress, as she refused to have any interference to reduce the fetal head size. The baby girl weighed 3.8 kg and the Apgar scores were 6 and 9 at one and five minutes, respectively. There were no anomalies except for the hydrocephalus.

A shunt operation was performed on the second day post delivery, but the baby died at six weeks of age due to chest infection. CT of the fetal brain confirmed bilateral dilatation of both ventricles with no other anomalies. Her chromosomal analysis was normal.

## Discussion

The presentation of this unusually high recurrence rate of familial CH in these two families is to highlight this problem. Consanguineous mating is a prevalent feature among the Saudis, as with other Arabs, and counseling for this congenital disorder is needed, especially as prenatal diagnoses can be advised for the affected families, and appropriate steps taken to encourage them against pregnancy. This is even more relevant when one considers the attendant childhood neuro-developmental disorders that may accompany such cases.

Hence, diagnostic methods for fetal hydrocephalus early in pregnancy, especially the X-linked type, could prove to be of immense benefit in deciding whether or not to terminate the pregnancy. Prenatal diagnosis has recently become available by means of chorionic villous sampling (CVS); X-linked hydrocephalus has been shown to carry a gene with mutation at the Xq28 which can be localized by linkage analysis or L1 mutation analysis [5]. Therefore, these cases can now be diagnosed in early pregnancy, and termination of pregnancy which could be accepted by Saudi families can be offered. In addition asymptomatic female carriers in suspected families can be detected. The autosomal recessive form has not been studied genetically but consanguinity rates of 96.5% among parents have been reported [3].

On the other hand, fetal ultrasonography has been reported to be useful for the early diagnosis of CH at 18 or 19 weeks of gestation in cases at risk for X-linked hydrocephalus. Serial sonographic monitoring of suspect fetuses every two to four weeks, starting at 16 weeks of gestation for high-risk pregnant women especially with

carrier status, should be a standard practice. However, prenatal sonographic diagnoses of affected male fetuses may still prove unreliable because the onset of hydrocephalus is said to be variable.

The long-term outlook for children with shunted hydrocephalus is said to be favorable in borderline non-progressive isolated ventriculomegaly. Indeed, in a cohort study of 155 hydrocephalic children with shunts, Casey *et al.* [6] reported 59% of them survived till school age and were able to attend a normal school. While the prognosis may be poor for the X-linked CH early diagnosis especially in cases of isolated CH may offer shunting facilities in order to avail the child a rare opportunity of relatively normal intellectual development.

The current availability of prenatal genetic diagnosis is another important issue to be discussed with such families especially for the X-linked cases. Such families are also good candidates for the study of biochemical screening markers in the first and second trimester of pregnancy for congenital hydrocephalus.

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