Sex linked hydrocephalus

SUMMARY A family showing the syndrome of X-linked hydrocephalus is presented and the relevance of this condition in genetic counselling discussed. A method of decompressing a grossly enlarged after-coming head is described.

Case history

A 29-year-old patient presented for hospital booking at 16 weeks' gestation by dates. Her only previous confinement, conducted elsewhere, had resulted in an intrapartum anoxic stillbirth after clinical evidence of fetal distress in early labour. A necropsy of this female infant (birthweight 3360g) had shown no evidence of congenital abnormality.

Examination confirmed the presence of a 16-week pregnancy. When seen at 32 weeks' gestation clinical examination revealed a breech presentation. This presentation was maintained and delivery by elective caesarean section was planned. An abdominal x-ray film, requested to exclude the possibility of gross fetal abnormality, showed an overlarge fetal head with thinning of the skull bones and widening of the cranial sutures. Sonar cephalometry at 36 weeks' gestation calculated a biparietal diameter of 11.4 cm confirming the diagnosis of hydrocephalus.

The patient was admitted to hospital at 38 weeks' gestation when a further sonar measurement showed an increase of 1.1 cm in the biparietal diameter. The findings were discussed with the patient and her husband and the reason for preferring vaginal delivery were explained. Labour was induced by low amniotomy and an escalating syntocinon infusion. Epidural analgesia was provided and, after a first stage lasting 6 hours, the patient reached full dilatation and was, therefore, encouraged to bear down with contractions. The breech was delivered easily but difficulty was experienced with the shoulders and general anaesthesia was induced. There was no associated spina bifida. The aftercoming head remained high in the abdomen and a wide bore needle was inserted per abdomen releasing approximately 600 ml cerebrospinal fluid. The head then entered the pelvic brim and an incision was made in the skin over the highest accessible part of the fetal neck; with blunt ended scissors, a 'tunnel' was then created upwards between the scalp and the underlying skull. A Simpson's perforator was inserted and, while steadying the fetal head with the left hand on the lower abdomen, the skull was perforated. The blades of the instrument were opened and rotated within the cranium releasing a further gush of cerebrospinal fluid and brain tissue. The collapsed head was then readily delivered without the use of forceps. The infant, a male, weighed 2860g. In view of the destruction of the skull contents necropsy was not requested.

The patient made an uncomplicated recovery and was discharged home two days later.

Family history

The patient's mother had had a stillborn hydrocephalic male infant in her first pregnancy in 1938, but no records remain of the details.

The patient stated that her elder sister had also had a stillborn hydrocephalic male infant in her first pregnancy. This was confirmed by reference to the hospital notes, which showed that hydrocephalus had been diagnosed by x-ray film one week before the onset of labour. During labour, with the vertex presenting, the head had to be perforated to allow delivery of the child. No necropsy had been requested because of damage to the brain and skull during the birth. Subsequently a normal daughter and son were born to this sister.

The family pedigree has been traced for three
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generations and is shown diagrammatically in the Fig.

Fig. The family tree of the case described. The carrier female, arrowed in the diagram, refers to the patient under discussion.

Discussion

The family history is compatible with an X-linked recessive lethal disorder, and on this hypothesis the patient, her sister, and her mother are all carriers of the gene responsible. The sister's daughter may prove to be a carrier.

Estimates of the incidence of congenital hydrocephalus, not associated with myelomeningocele, vary from 0.22 per thousand live births to 1.8 per thousand total births. It appears to be unusual for this uncomplicated form of hydrocephalus to present in more than one member of the family. However, Bickers and Adams (1949) reported a family in which a woman had 3 sons and 4 brothers all dying at birth from hydrocephalus. Necropsy of one of the affected infants showed pronounced narrowing of the aqueduct of Sylvius. Edwards et al. (1961) reported an even larger family with no fewer than 15 affected males, whose distribution within the family pedigree suggested an X-linked recessive mode of inheritance. Necropsy of the index patient showed aqueductal stenosis.

Further cases have been reported by Warren et al. (1963), Karim et al. (1964), and Shannon and Nadler (1968), all confirming the syndrome of congenital hydrocephalus caused by stenosis of the aqueduct as an X-linked recessive disorder. Those affected are usually stillborn; infants born alive may show severe mental defect, a variable degree of hydrocephalus, opposed thumbs, and spasticity of the lower limbs. A leading article (British Medical Journal, 1962) considered that the syndrome could account for perhaps 2% of all cases of uncomplicated hydrocephalus.

The existence of this syndrome, like others of which hydrocephalus is a feature, should be remembered when counselling parents who have had one hydrocephalic male infant without evidence of spina bifida. In the X-linked condition, where a mother is known from the family history to be a carrier, the recurrence risks will be 50% for any son and, though none of her daughters will be affected, 50% will in turn be carriers. At present there is no way of identifying the carrier status.

Estimation of the alphafetoprotein level in the amniotic fluid in early pregnancy is unhelpful in cases of hydrocephalus not associated with spina bifida, since there is no communication between the cerebrospinal and amniotic fluids. However, today antenatal amniocentesis and cell culture to identify the fetal sex can be undertaken and selective abortion of male fetuses considered (though on average 50% of these would be normal). This procedure would possibly lead to an increase in the absolute number of female carriers and a corresponding increase in the number of pregnancies at risk. It is very doubtful indeed if such a practice would provide a real answer either to the individual or the community. Armed with the above information, our patient has decided to avoid further pregnancies and adopt a child.

References


