and the pregnancies of her daughters (III.4 and 5) in
the more distant future.

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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 3360 g) 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 diam-
eter. The findings were discussed with the patient
and her husband and the reason for preferring
vaginal delivery were explained. Labour was in-
duced by low amniotomy and an escalating syntoci-
on 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 shoul-
ders 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 approxi-
ately 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 under-
lying 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 cere-
brosplinal fluid and brain tissue. The collapsed head
was then readily delivered without the use of forceps.
The infant, a male, weighed 2860 g. 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 hydroce-
phalic 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 re-
quested 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
Case reports

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.

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