RES MEDICA
Journal of the Royal Medical Society

Congenial Maldevelopments as a Cause of Hydrocephalus

David Doyle

Abstract
The incidence of congenital malformation is difficult to gauge accurately. In Philadelphia, in the late 1930's, 2.9% of 7,478 cases of still-birth had malformations recorded on the death certificates. About the same time, a rate of 20..2% was derived from autopsy reports on 435 still-births in Edinburgh. The percentage of children who died in the neonatal period with abnormalities varied between 10.5% in Edinburgh, in Glasgow and 13% in Belfast. There has been no increase in the incidence of congenital defect recently.

Hydrocephalus comes second to anencephaly as a cause of death from congenital abnormality.

The rarity of malformations causing hydrocephalus makes elucidation of aetiology difficult. but significant correlation has been found in some instances.

ISSN: 2051-7580 (Online) ISSN: 0482-3206 (Print)
Res Medica is published by the Royal Medical Society, 5/5 Bristo Square, Edinburgh, EH8 9AL

Res Medica, Summer 1960, 2(2): 32-35
doi:10.2218/resmedica.v2i2.340

Congenital Maldevelopments as a Cause of Hydrocephalus

By DAVID DOYLE

Based on a Dissertation read before the Royal Medical Society on Friday, 13th October 1959.

(i) Incidence and Aetiology

The incidence of congenital malformation is difficult to gauge accurately. In Philadelphia, in the late 1930's, 2.9% of 7,478 cases of still-birth had malformations recorded on the death certificates. About the same time, a rate of 20·2% was derived from autopsy reports on 435 still-births in Edinburgh. The percentage of children who died in the neonatal period with abnormalities varied between 10·5% in Edinburgh, 2·5% in Glasgow and 13% in Belfast. There has been no increase in the incidence of congenital defect recently.

Hydrocephalus comes second to anencephaly as a cause of death from congenital abnormality.

The rarity of malformations causing hydrocephalus makes elucidation of aetiology difficult, but significant correlation has been found in some instances.

Much less clearly than with mongolism, the increased proportion of hydrocephalics born to mothers over 35 years of age has been noted. Between 1940-47 the incidence among children born to mothers in the 40-49 age groups was three times greater than among those of mothers under 30. Another series in 1947 showed that from mothers over 40 years the incidence was twice that from the under 30 group. It is not possible to dissociate birth rank from maternal age, because an increase only becomes apparent after the fifth or sixth pregnancy.

Congenital hydrocephalus shows no obvious distribution towards either sex, although one series showed a male : female ratio of 1·27 : 1. Hydramnios, and less often oligohydramnios, has sometimes been shown to be associated with hydrocephalic infants.

Lack of Vit. A increases the proportion of hydrocephalic young born to rabbits and, in rats, deficiency of Folic acid and Vit. B12 can be made to produce almost pure families with deformed aqueducts and hydrocephalus.

The high mortality rate from hydrocephalus does not lend itself to studies of hereditary trends, but some observations have been made. Mendelian recessive traits led to narrowing of the aqueduct in one family and retarded cartilage growth at the base of the skull in another. Obstruction of the Foramen of Monro is said to be hereditary.

More than one-third of congenitally hydrocephalic children present some other congenital malformation.

(ii) Pathology

Most of the errors of development occur at points in the cerebro-spinal

fluid channels where there is a constriction. They may represent failures of opening, delays in opening, or reclosing of a space.

Aqueduct of Sylvius—	Stenosis	(fairly common)
	Complex Forking	(common)
	Gliosis	(rare)
	Septae	(very rare)
Foramina of Magendie and Luschka—	Blockage	(common)
	Failure to open	(rare)
Dandy-Walker Syndrome—		(rare)
Arnold-Chiari Syndrome—		(very common)
Lissencephaly		(very rare)
Deformities of Skull—	Platybasia	(rare)
	Achondroplasia	(rare)
	Defects of Sutures	(rare)

Aqueduct of Sylvius

Stenosis is the least common aqueduct disorder causing hydrocephalus. Enormous variations in the diameter of the tube are encountered, and the foetal aqueduct is wider than the adult's. There are usually two constrictions in the aqueduct, one—0·4-0·8 sq. mm.—at the level of the superior colliculus, and another—0·5-1·0 sq. mm.—at the level of the intercollicular sulcus. With the merest pin-point of an aqueduct, people may live without trouble, others develop attacks of headache, vertigo, nausea and vomiting, while others may become frankly hydrocephalic.

Forking may be simple, in which case it is harmless and common, or complex, a common or certain cause of hydrocephalus. Many normal individuals have outpouchings from the dorsal, lateral or more commonly ventral walls of the aqueduct. These "simple forks" are blind tubes, not interfering with the patency of the main tube. In complex forking, on the other hand, both tubes divide repeatedly, establishing ineffective or no communication with the rest of the duct. The flow of C.S.F. from the third ventricle is stopped.

The branches of the aqueduct are separated by normal neural tissue. The corpora quadrigemina and third nerve nuclei are often fused. These facts have been taken to illustrate an exaggerated closure of the neural tube in this region.

Gliosis of the Aqueduct is uncommon. It often looks like a neoplasm and, since normal people often have rests of ependymal cells in the surrounding neural tissue, it could be a neoplasm of these cells. Basing their argument on the widespread ependymal disorder which often occurs with gliosis, some hold this to be part of the aftermath of a foetal ependymitis.

Gliosis is common in cerebral diplegia, with and without hydrocephalus.

Septae of neuroglia across the aqueduct have been reported.

Foramina of Magendie and Luschka

The Foramina of Magendie and Luschka may be imperforate at birth, or may have been closed again by toxoplasmosis or congenital syphilis. Even if the foramina do not break through, severe degrees of hydrocephalus are not the rule; sufficient fluid is able to cross the membrane under ordinary conditions to maintain a balance compatible with adult life in many instances. These subjects, if not hydrocephalic, show a tendency to sudden pressure elevation caused by such diverse factors as physical exertion, alcoholic bouts, fatigue and menstruation. They are, of course, poorly placed should they develop a serous meningitis.

When the foramina are absent, defects of the medulla and cerebellum are common.

The Dandy-Walker syndrome is one in which there is hydrocephalus with deficiencies of the cerebellar vermis. In the embryo, when the cerebellar anlages migrate across the roof of the fourth ventricle, they fuse anteriorly first. Stretching of the roof membrane might delay fusion of the posterior parts. If bulging, caused by raised C.S.F. pressure, is the cause of the cerebellar defect, the C.S.F. production must begin inordinately early, because the fusion of the anlages is complete before the foramina are due to open. Their opening is related in time to the beginning of function of the choroid plexuses at the fifth month of intra-uterine life. This syndrome is easy to recognise. The occuput is long; the lateral sinuses and tentorium are elevated; ventriculography is distinctive. Fortunately, it is especially amenable to treatment.

The Arnold-Chiari syndrome of hydrocephalus with cerebellar displacement and meningocoele is a common cause of hydrocephalus. About 20% of hydrocephalic infants have meningocoeles and, if one appears at a breech presentation, a hydrocephalic child must be expected.

Features of this syndrome are fusion of the cerebellar tonsils and their protrusion downwards through the foramen magnum over a long, lower brain stem to the upper cervical cord. The upper cervical roots are angled upwards, and the medulla inclined backwards on the cord. There is invariably a major degree of spina bifida, and two-thirds of all causes have microgyria.

The cerebellar tissue involved in the deformity is hypoplastic, and the small cerebellum lies in a small posterior fossa—craniolacunia. The cerebellar abnormality does not seem to contribute to the hydrocephalus. The smallness of the posterior fossa seems to cause enlargement of the posterior horns of the lateral ventricles.

The leptomeningeal vessels—especially, but not entirely, the veins overlying the cerebellar deformity are increased in number and size. Half of all cases have abnormal vessels in the mid-brain.

The causes of hydrocephalus in this syndrome are complex forking of the aqueduct in some, and probably hindbrain compression because of the smallness of the posterior fossa. Some may have impaction of the foramen magnum because of the cerebellar and vascular structure around the opening. A contributing factor may be that the exit foramina of the fourth ventricle are below the foramen magnum.

Why these abnormalities should occur in such constant partnership is not known. Lichtenstein's theory of traction from below is untenable because, even without a fixed meningocoele, upward angling of the cervical roots may occur. This angling occurs in the Klippel-Feil syndrome of congenital short neck. One theory is that there has been pressure on the foetal head during intra-uterine life, squeezing the hind-brain through the foramen magnum. There is, however, no relation between the amount of basilar impression and the degree of cerebellar displacement.

There are seldom diagnostic clinical features of this condition. Cerebellar signs, such as mirror movements of the arms, unsteadiness, clumsiness and nystagmus are common, but not constant. Intermittent pressure increases are common in children and adults, bringing severe headaches—often precipitated or aggravated by coughing or sneezing—tinnitus, nausea and neck stiffness.

That the meningocoele may maintain the balance between absorption and production of C.S.F. is debatable. Some series show that upwards

of 15% of those who have had them removed develop increased pressure. The sac may participate in absorption, but it may not be assumed that the removal of the sac alone causes pressure elevation.

Lissencephaly, or agenesis of the subarachnoid space, which normally opens after ten weeks of intra-uterine life, is added for completeness.

Another rare anomalous cause consists in large, detached masses of tissue lying free in the meninges. This is usually associated with neoplasia elsewhere.

Malformations of the skull predisposing to hydrocephalus are rare.

In **Platybasia,** the angle between the basi-sphenoid and the basi-occiput is increased beyond the normal 130-140 degrees. The posterior fossa is foreshortened, the margins of the foramen magnum become inverted, and the odontoid process, appearing in the posterior fossa, impinges on the hindbrain. This occurs as a congenital defect, or may appear as a result of modelling of soft bone, as in osteitis deformans.

Achondroplasia, by altering the relations at the base of the skull, causes hydrocephalus.

Craniostenosis and **Oxycephaly,** conditions of abnormal fusion of sutures, reduce the capacity of the skull, and occasionally show elevated C.S.F. pressure.

REFERENCES

1. Dorothy Russell, "Observations on the Pathology of Hydrocephalus." Special Report Series, M.R.C., 1949.
2. Biggart, "Pathology of the Nervous System." 2nd Edition. 1949.
3. "Modern Trends in Neurology." Second Series. 1957.
4. J. D. Ballantyne, "Antenatal Pathology: Infant." 1904.
5. A. MacFarlane and A. J. F. Maloney, "The Appearance of the Aqueduct and its Relationship to Hydrocephalus in the Arnold Chiari Syndrome." "Brain," 1957.
6. R. A. Willis, "The Borderlands of Embryology and Pathology." 1958.
7. J. E. Morrison, "Foetal and Neonatal Pathology." 1952.
8. A. D. Bain, "Perinatal Pathology." M.D. Thesis. Edinburgh.
9. A. Brodal and E. Haughlie-Hanssen, "Congenital Hydrocephalus with Defective Development of the Cerebellar Vermis—Dandy Walker Syndrome." "J. Neurol. Neurosurg. Psychiat." 1959.
10. J. Frazer and N. Dott, "Hydrocephalus." B.J.S. 1922-23.
11. M. Gordon, "Craniostenosis." B.M.J. October 24, 1959.