Acta Neuropathol (Berl) (1984) 63:330–333

Acta Neuropathologica

Congenital Ventricular Diverticulum in the Brainstem

Report of Four Cases*

A. Hori[1], R. Eubel[2], and R. Ulbrich[3]

Depts. of [1] Neuropathology, [2] Human Genetics, and [3] Gynecology and Obstetrics, University of Göttingen, Robert-Koch-Str. 40, D-3400 Göttingen, Federal Republic of Germany

Summary. This report concerns rare ventricular malformations observed in four fetal brains. Multiple aqueductal diverticula were the only malformation of the central nervous system (CNS) in one case, unilateral diverticula of the 4th ventricle in a holoprosencephaly and a hydrocephaly, and a ventral accessory aqueduct in a fetus with trisomy 18. These findings are the pathologic progression of embryonal ventricular recesses which may be seen in normal brains.

Key words: Aqueduct of Sylvius – 4th ventricle – Malformation of ventricular system – Ventricular diverticulum

Introduction

There are few observations on malformations of the ventricular system. After finding diverticula of the lateral ventricle in a case of cloverleaf skull anomaly (Hori et al. 1983), we found another four cases of congenital malformations of the aqueduct and 4th ventricle. We present these cases and discuss the pathogenetic mechanisms.

Case Reports

Case 1 (A 200/81)

A male fetus with a 14-cm crown-heel length (11.5 cm crown-rump length) was born to a 33-year-old mother who had previously had three spontaneous abortions due to unknown cause. She had no live birth.

General examination of the stillborn and the placenta showed no pathologic changes.

The brain was macroscopically normal. The brain was cut serially. The only abnormalities found were the absence of the massa intermedia and changes of the aqueduct. There was a slit-like dorsal elongation of the aqueduct (A 1 in Fig. 1a, b) close to its origin. The ependymal cells in the diverticulum A 1 were high columnar in contrast to cuboidal cells in the normal aqueduct. No structure suggesting a subcommissural organ was seen in the region. At the level of the rostral superior colliculus, a second recess (A 2 in Fig. 1a, b) appeared ventrally to the aqueduct extending in a ventral direction. Another outpouching was found dorsally in the midline, extending for 1.4 mm (A 3 in Fig. 1a, d). The fourth sac-like structure was found on the left side at the beginning of the 4th ventricle extending into the pontine tegmentum (A 4 in Fig. 1a, e, f). The remainder of the 4th ventricle was normal.

Case 2 (A 332/83)

A female with craniofacial anomalies (short and steep forehead, microphthalmia, blepharophimosis, and single opening of the nasal cavity) was born to a 30-year-old primigravida. The pregnancy was uneventful up to the 18th gestational week, when "hydrocephalus" was diagnosed sonographically. The mother delivered at home. The infant was apparently asphyxiated, with Apgar's scores of 0/1/0, and died during transport to the hospital.

The cause of death was amniotic aspiration; the only visceral anomaly was a bicornuate uterus.

The brain showed typical holoprosencephaly with olfactory aplasia (Fig. 2a) and aplasia of the falx and hypoplasia of the tentorium. In the floor of the 4th ventricle, there was a unilateral diverticulum at the level of the abducens nucleus on the right side (Fig. 2b). The recess was surrounded by hyperplastic subependymal tissue. The foramina of Luschka were in the normal position.

Case 3 (A 436/83)

Thirty-nine-year-old mother with one healthy child had an uneventful pregnancy until a unilateral hydrocephalus of the fetus was diagnosed by ultrasound in the 26th gestational week (Fig. 3a). Labor was induced by prostaglandin. The male fetus, 35 cm long with a head circumference of 27 cm, showed no external anomalies.

The necropsy was performed 4 days after death; the brain was autolytic. The telencephalon showed normal gyration for the age. There was bilateral hydrocephalus, the right lateral ventricle being larger than the left. Due to autolysis, the examination of the telencephalon was limited. A small recent subependymal hemorrhage was seen on the right. There was no atresia of the aqueduct. Histological study revealed a unilateral diverticulum in the bottom of the 4th ventricle, lateral to the right abducens nucleus (Fig. 3b).

* Dedicated to Professor Dr. Hans Orthner on the occasion of his 70th birthday on August 4, 1984

Offprint requests to: Dr. A. Hori (address see above)

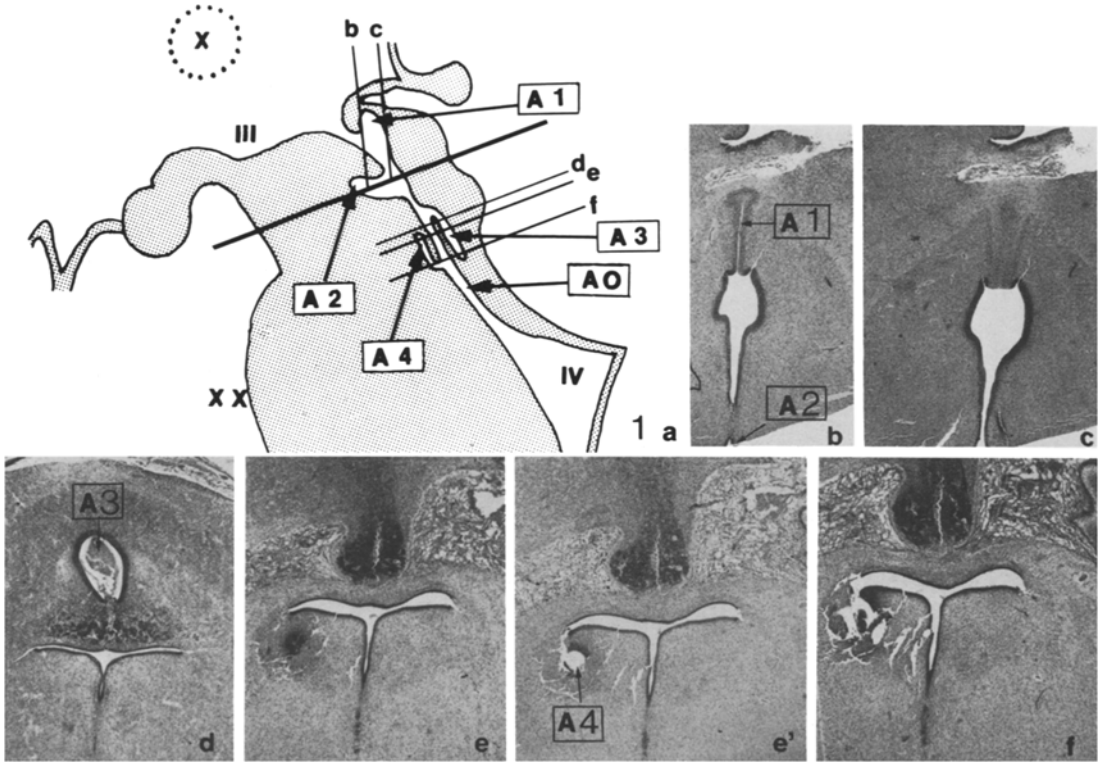

Fig. 1a—f. Case 1. Multiple diverticula of the aqueduct. **a** Schematic drawing of diverticula; *AO*, aqueduct of Sylvius; *A1—A4*, diverticula; *III*, 3rd ventricle; *VI*, 4th ventricle; *X*, absent massa intermedia; *XX*, physiologic hypoplasia of the pons. **b—f** Histology of the diverticula from serial sections. The corresponding planes of cut are indicated in **a. b—e** Nissl stain, ×12.5; **f** HE, ×10.6

Case 4 (A 268/81)

Trisomy 18 and marked elevation of alpha-fetoprotein (72,000 IU) were revealed by amniocentesis in the 16th gestational week of the 43-year-old para II, gravida III. The female fetus, 12 cm long, was stillborn in the 22nd gestational week and showed sacral spina befida and rocker bottom feet. No other malformations were found except a small aberrant accessory aqueduct between the oculomotor nuclei. Dorsal to the normal aqueduct there were aggregations of glial cells suggesting the rudimentary remnant of recessus mesocoelicus (Fig. 4).

Discussion

The pathogenesis of these anomalies should be analyzed in the light of the embryology of the aqueduct and the 4th ventricle as described by Turkewitsch (1935) and the normal variations of the surface structures of these regions as described by Friede (1961). A persistent recessus mesocoelicus (or anterior recessus mesencephali) is found on the dorsal aspect of the aqueduct close to its origin (Fig. 5,2). On the caudal dorsal aspect of the aqueduct, there is a remnant of the posterior recessus mesencephali (Fig. 5,6). The floor of the aqueduct may show variable forkings (Fig. 5,5). In an embryo at 8 cm crown-rump length stage, the recessus isthmi is observed in the ventroposterior part of the aqueduct. It becomes shallower and shifts oralward during subsequent growth to 21 cm of crown-rump length (Turkewitsch 1935).

The diverticulum A 1 in case 1 may be due to a pathologic enlargement or failure of closure of the recessus mesocoelicus. The diverticulum A 3 could have similar relationship to the posterior recessus mesencephali. The diverticulum A 2 can be compared with the embryonal recessus isthmi.

The unilateral basal diverticulum in cases 1 (A 4), 2, and 3 may be explained as a result of pathologic outpouching of the furrowed area of the aqueduct and the 4th ventricle. The diverticulum of case 2 could also result from hyperplasia of the neighboring subependymal tissue. The site and morphology of the diverticulum, however, were identical to that in case 3 which did not show hyperplasia of the subependymal tissue. Therefore, both diverticula are similar, although they occurred in two different malformtions. In case 3, the cause of the hydrocephaly remained unclear because of advanced autolysis of the brain; no aqueductal atresia was found.

According to Friede (1961), the forking of the floor of the aqueduct results from incomplete fusion of the median fissure and is biologically not significant.

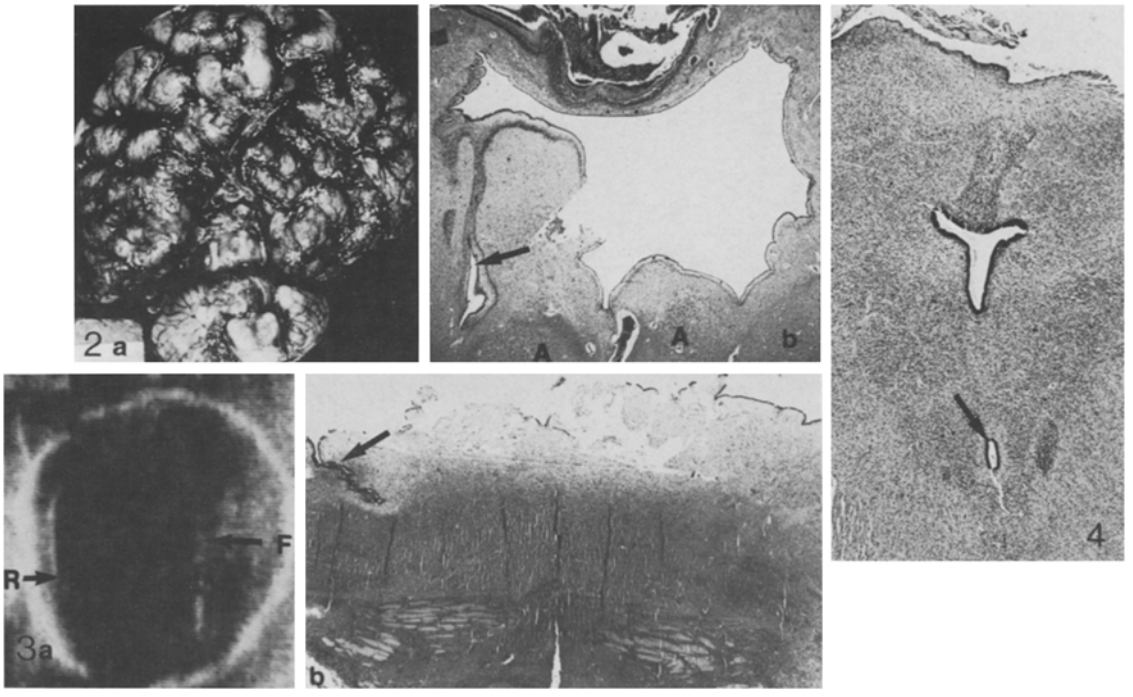

Fig. 2a, b. Case 2. **a** Ventral view of the brain; **b** unilateral diverticulum of the 4th ventricle with local hypertrophy of the subependymal tissue at the level of the abducens nuclei (**A**). HE, ×7.7

Fig. 3a, b. Case 3. **a** Ultrasonic picture cut horizontally; asymmetric enlargment of lateral ventricles. *F*, falx; *R*, right lateral ventricle. **b** Unilateral diverticulum of the base of the 4th ventricle. HE, ×7.7

Fig. 4. Case 4. Ventral accessory aqueduct at the level of the oculomotor nucleus. HE, ×13.1

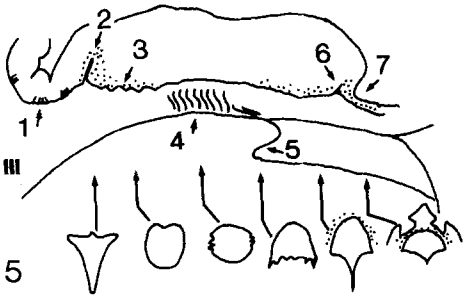

Fig. 5. Schematic drawing of surface structures in the human aqueduct as modified from Friede (1961). Recessus mesocoelicus (*2*), ventral forking of aqueduct (*5*), and remnant of recessus mesencephalicus posterior (*6*) are indicated

However, both dorsal and ventral forkings are not common. In case 4, trisomy 18 was documented. Neuropathologic changes in trisomy 18 are well documented (Terplan et al. 1966; Sumi 1970; Michaelson and Gilles 1972); however, no description of a ventricular anomaly is found in the literature. The "complex (dorsal and ventral) forking" with two distinct channels in the mid-sagittal plane was observed in a case of Arnold-Chiari malformation by MacFarlane and Maloney (1957). Ventricular anomalies are usually associated with other brain malformations and the dorsal forking of the aqueduct (case 1), together with other aqueduct anomalies, has not been reported in otherwise normal individuals.

The massa intermedia is absent in 3% of brains between 17 weeks gestation and 19 years after birth as a result of secondary loss (Lemire et al. 1975). In our case 1, its absence may be due to aplasia, associated with aqueductal anomalies.

A similar diverticular condition in the mesencephalon has been recorded in three adult cases (Spiller 1906; Hardy and Stevenson 1957; Samples et al. 1983). The reports concern a "syrinx", partially lined by ependyma (in the latter two reports). Samples et al. (1983) stated that it was probably an unusual diverticulum of the aqueduct.

The term "ventricular diverticulum", as used by neurosurgeons and radiologists, means an acquired or secondary diverticulum caused by rupture of the lateral ventricle, generally at the level of the medial wall of the trigone, or of the 3rd ventricle at the level of the supra- or infra-pineal recessus (Alonso et al. 1979; Kapila et al. 1981; Wakai et al. 1983). The congenital ventricular diverticula should be distinguished clearly from the acquired changes.

Acknowledgement. We are grateful to Professor R. L. Friede and Professor U. Roessmann for their useful suggestions and constructive criticism.

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Received January 13, 1984/Accepted February 13, 1984