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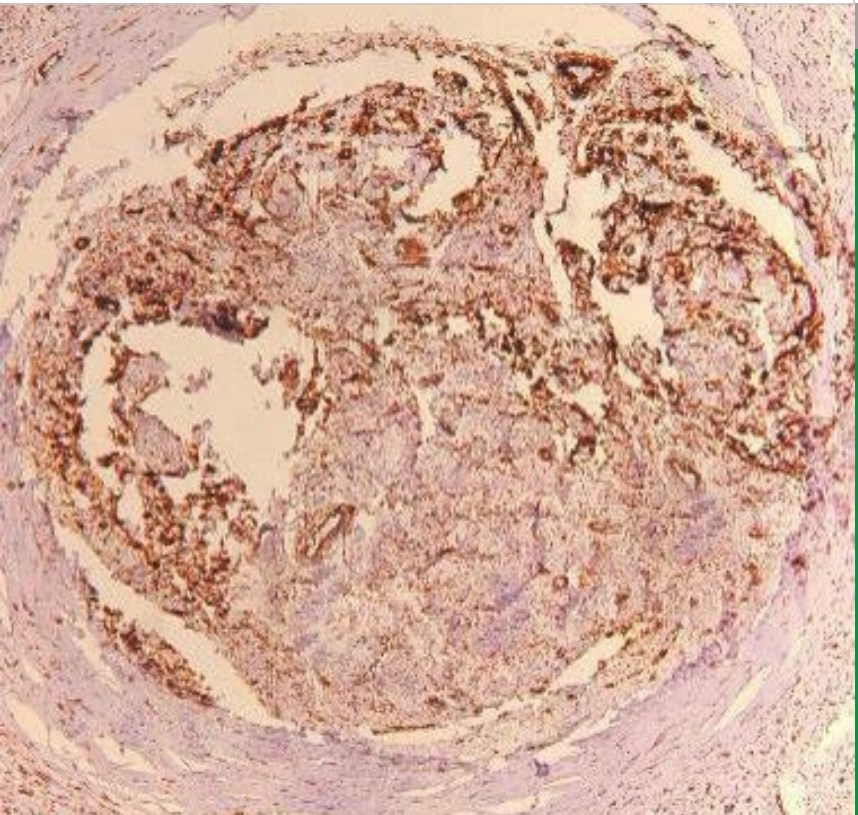
Hydranencephaly or mid-gestational disruption of brain perfusion



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A 22-year-old G1/P1 was admitted at 38 1/7 weeks of gestation with spontaneous rupture of membranes. There was no prenatal care. On admission, fetal ultrasound revealed a severe cerebral malformation diagnosed as hydranencephaly. Maternal serologies were normal. Head circumference was estimated at 37 cm (> P90). Because of the infant's poor prognosis, 500 ml of cerebral fluid were aspirated to facilitate vaginal delivery, and the infant was born 24 hours later.

Initially, the infant was bradycardic without spontaneous breathing and poorly reactive. Apgar scores at 1 and 5 minutes were 5. Without any stimulation, the baby began to breathe at 6 minutes of life and the heart rate increased above 100/min. At 10 minutes, the Apgar score was 9.

Birth weight was 2420 g (< P10), length 45 cm (< P10), and head circumference 34 cm (P50-90). The physical examination was unremarkable with a normal appearing head. The diagnosis of hydranencephaly was confirmed by ultrasound and MRI (Fig. 1). Abdominal ultrasound was normal and karyotyping revealed no chromosomal anomaly.

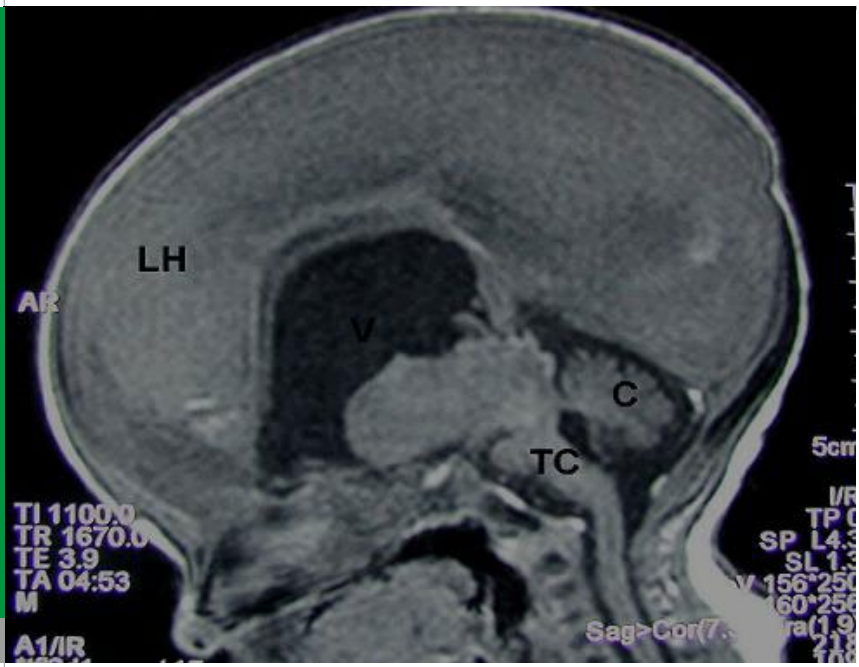

Fig. 1

MRI (left paramedian sagittal section): V: enlarged 3rd and lateral ventricles in continuity, forming one large ventricle; C: Cerebellum; LH: Cavity corresponding to left hemisphere; TC: Brain stem

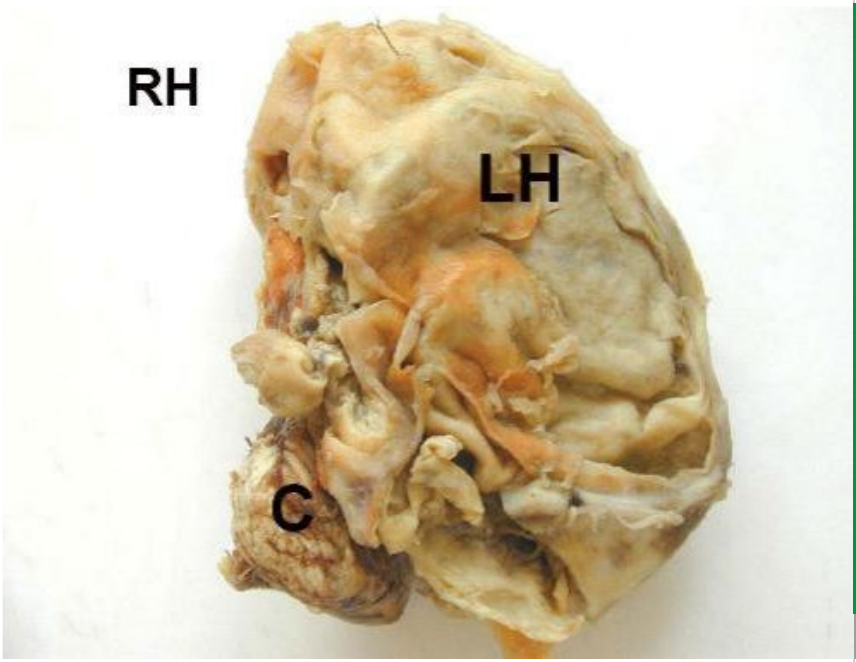

Fig. 2

Autopsy findings (after fixation): LH: left hemisphere; RH: right hemisphere; C: cerebellum

The baby was fed through a nasogastric tube. He developed a periodic breathing pattern with severe apnea spells of long duration. To comfort the baby, sedation with morphine was introduced and later complemented with diazepam. The baby died at 5 weeks of age.

Autopsy confirmed the diagnosis of a severe cerebral defect with extensive destruction of the cerebral hemispheres, mainly in the territories of the anterior and the middle cerebral arteries. The temporal, occipital as well as a small part of the frontal lobes were still discernible. The deep grey nuclei were partly visible. The brainstem and cerebellum were only focally involved (Fig. 2).

On histology, there was a significant disarray of the cytoarchitecture with formation of polymicrogyri suggesting an early insult during the first part of the second trimester. The most severely affected regions showed fibrosis, numerous hemosiderin-laden macrophages and gliosis. The circle of Willis as well as the internal carotid arteries were widely patent. Only a small vessel in the surrounding fibrotic arachnoid showed fibrous obliteration of its lumen with little neovascularisation (Fig. 3).

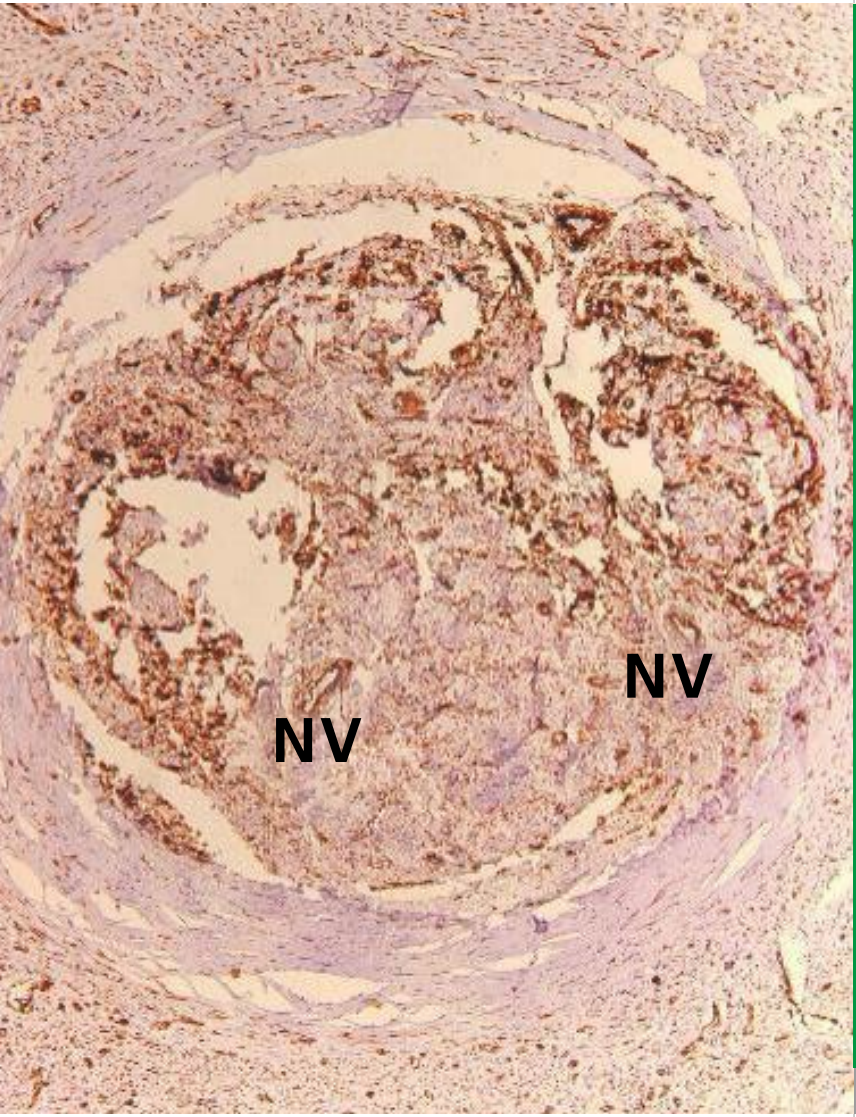

Fig. 3

Immunhistochemistry (CD 34): Obliterated arachnoidal vessel with secondary neovascularisation. NV: secondary formed recanalizing vessels.

DISCUSSION

Hydranencephaly is a rare condition defined as extensive destruction of the brain caused by disruption of brain perfusion. The normal brain tissue is replaced by a membranous sac, which is filled with fluid (1). In a series of 4122 autopsies, 363 cases revealed some congenital CNS malformation (235 liveborn infants and 128 stillborn infants), but hydranencephaly was diagnosed in only two patients (2).

Disruptions affect mainly the major vessels of the anterior and middle cerebral arteries. Up to the 20th week of gestation neuronal migration is still active. The earlier disruptions occur in the second trimester, the more migrational disorganisation may be found. After 20 weeks of gestation, the brain lesions are not accompanied by cytoarchitectural disorders. During this midgestational period, the neural tube is already closed and well developed. Intact skull and scalp are covering the underlying central nervous system structures and the head appears normal.

Fetal cerebral perfusion can be affected by A) maternal conditions (cardiovascular collapse; trauma; gas poisoning; coagulopathy; cocaine abuse), B) fetal conditions (multiple pregnancy; non-immune hydrops fetalis; blood dyscrasia), or C) placenta and cord pathologies (infarct; calcifications; knot of the cord; neoplasm) (3). In surviving infants, neuromotor activity is very limited with some patients developing uncoordinated movements. Convulsions are rarely descri-

bed because of the absence of a motor cortex in most patients. The sensory capacities are also very limited with some reaction to strong light. In very rare cases, crying and smiling may be seen. Prolonged survival of up to 19 years can occur with hydranencephaly; it is, however, not associated with any improvements in consciousness or awareness (4).

Electrophysiologic features cannot be used as predictors of the length of survival, whereas some authors have suggested a functioning hypothalamic-pituitary-adrenal axis to be essential for prolonged survival.

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