Familial communicating hydrocephalus, posterior cerebellar agenesis, mega cisterna magna, and port-wine nevi

Report on five members of one family

HARVEY R. NOVA, M.D.
Surgical Service (Neurological Surgery), Englewood Hospital, Englewood, New Jersey

This report deals with a family in which the maternal grandmother, the mother, and all three male children have port-wine nevi and mega cisterna magna. Two of the three male children have, in addition, congenital communicating hydrocephalus associated with agenesis of the posterior cerebellar vermis. This case of familial communicating hydrocephalus, posterior cerebellar vermis agenesis, port-wine nevus, and mega cisterna magna represents a new neurocutaneous syndrome, possibly transmitted as an autosomal dominant.

KEY WORDS - hydrocephalus - vermis agenesis - neurocutaneous syndrome - autosomal dominant - cisterna magna

FAMILIAL, sex-linked recessive aqueductal stenosis with or without other anomalies is a rare, but well reported syndrome.4,7-9,18,17 This report presents two brothers with a syndrome consisting of glabella port-wine nevi, agenesis of the posterior cerebellar vermis (APCV), mega cisterna magna (MCM), and congenital communicating hydrocephalus. Study of the additional unaffected middle male child, the parents, and two grandparents by computerized tomography (CT) shows the same MCM in the male sibling, the maternal grandmother, and the mother, who also have glabella port-wine nevi. This is the first case report of a new neurocutaneous syndrome consisting of port-wine nevus, mega cisterna magna, and communicating hydrocephalus in association with posterior cerebellar vermis agenesis.

Case Reports

Case 1

This 9.5-month-old baby boy presented with a 3-day history of vomiting and lethargy. He was the product of an unmarred pregnancy and an uncomplicated delivery. His head circumference at birth and his developmental milestones up to the time of admission were normal. On admission, he had a head circumference of 45.5 cm. He was poorly responsive and had head retraction. He also had a glabella port-wine nevus. Cerebrospinal fluid (CSF) tap showed a protein of 150 mg/dl, and a normal sugar and cell count. Opening pressure was not measured. Nuclear brain scan and skull films were normal. All of the symptoms cleared and he was discharged with a diagnosis of possible viral encephalitis.

Two months later the patient returned with an increase in head circumference of 7.5 cm, bilateral lower extremity spasticity, and bilateral ankle clonus. Pneumoencephalogram and ventriculogram showed air entering a mega cisterna magna, agenesis of the posterior cerebellar vermis, a dilated ventricular system, along with a dilated fourth ventricle and an elevated tentorium (Fig. 1). A minimal amount of air passed distally into the basal cisterns. The ventricular and spinal fluid contained 117 mg/dl and 113 mg/dl of protein, respectively. A ventriculooatrial shunt was
FIG. 1. Case 1. The combined pneumoencephalogram-ventriculogram shows the mega cisterna magna and the elevated tentorium-torcular complex (horizontal arrow). The dilated fourth ventricle is also shown (vertical arrow).

inserted, but removed because of sepsis; reinsertion was successful after antibiotic therapy. Work-up for infectious meningitis was negative.

The patient has returned for multiple ventricular catheter revisions. Temporal lobe seizures developed when he was 7 years old and have been controlled with phenobarbital. Intellectually, he is functioning above his age level with only some difficulty in spatial relationships. A CT scan at 9 years old confirmed the findings on air study (Fig. 2).

Case 2

The younger brother of the above patient was born after an uncomplicated pregnancy and delivery. His head circumference at birth was 36 cm. He presented at age 2.5 months with an increase in head circumference of 9.5 cm, and signs of increased intracranial pressure. A CT scan showed MCM, APCV, and a dilated ventricular system (Fig. 2). He, too, had a glabella port-wine nevus. A ventriculoperitoneal shunt was placed, and he has developed normally to the present time.

Cases 3, 4, and 5

The maternal grandmother, the mother, and the unaffected 8-year-old male child all have glabella port-wine nevi and MCM visible on CT scan (Fig. 2). The father and the paternal grandfather have normal CT scans and no skin lesions.

Discussion

The full syndrome presented by the two affected male children consists of glabella port-wine stain, APCV, MCM, and communicating hydrocephalus. The mother, the maternal grandmother, and the unaffected male child have only the port-wine nevus and MCM, representing the milder form of the syndrome.

The maternal grandmother had two siblings, the paternal grandfather three siblings, and the father four siblings, all of whom were normal. The pattern of inheritance for the complete syndrome may be that of an autosomal dominant with incomplete expressivity in the mother and the grandmother. This is different from the pattern of sex-linked recessive transmission found in x-linked aqueductal stenosis.4,7,9,13,15,17

The combined pneumoencephalogram-ventriculogram performed on Case 1 demonstrated the CSF dynamics well. Air injected via the lumbar sac or the ventricle, freely entered the dilated MCM and the dilated ventricular system, and showed their patency and their communication with each other. Air escaped and flowed poorly from the MCM into the basal cisterns. From here little air passed distally. This establishes the diagnosis of a communicating hydrocephalus with a block distal to the cisterna magna. The air study also showed the APCV and the undescended tentorium. In Case 2, the CT scan showed APCV and free communication between the fourth ventricle and the MCM (Fig. 2). Again the block is distal to the MCM. The youngest and oldest siblings have identical CT scans, allowing for their age differences, and for the fact that the oldest sibling had been shunted at the time of the CT scan.

These anomalies in the hind brain must occur before the third or fourth month of gestation, as this is the time of the fusion of the posterior cerebellar vermis.3 Corroborating evidence is seen in the presence of the elevated tentorium, which would normally descend at the fourth gestational month. Whatever genetic disturbance is responsible for this syndrome causes failure of fusion of the posterior cerebellar vermis, leaves the exit foramina of the fourth ventricle unaffected, but blocks the egress of CSF from the basal cisterns. Further dilation of the congenitally large cisterna magna then prevents egress of CSF from the basal cisterns. The hydrocephalus is incomplete at birth since the patients in both Cases 1 and 2 had normal head circumferences at birth. Partial temporary compensation for the communicating hydrocephalus is accomplished by egress of CSF from the cisterna magna into the spinal canal with its resultant absorption. This was confirmed by the unobstructed flow of air from the lumbar sac into the dilated cisterna magna. Absorption via the lumbar sac is obviously not sufficient for full long-term compensation, and with time the hydrocephalus becomes manifest.

The severely affected two male children show some of the radiological criteria described in the Dandy-
FIG. 2. A computerized tomography family tree. The maternal grandmother (A), the mother (B), and the children (C', C, C") all show the mega cisterna magna (long arrows). These are also the individuals with the port-wine nevus. The agenesis of the posterior cerebellar vermis is shown in C' and C" (short arrows). Hydrocephalus is seen in C' before shunting.

Walker syndrome, namely, gross enlargement of the posterior fossa and a high position of the tentorium and torcular. In this instance it is due to obstruction of the basal cisterns and not to fourth ventricular dilatation. These two children exhibit an appearance differing from that reported in infratentorial cysts, which are arachnoid cysts originating from the back of the fourth ventricle causing obstruction to flow from that ventricle. In the Dandy-Walker syndrome, there is generally complete obliteration of the cisterna magna, and in the infratentorial cysts there is no communication between the cisterna magna and the fourth ventricle. In our cases, the cisterna magna was dilated and there was free communication between the fourth ventricle and the cisterna magna.

Two other isolated nonfamilial cases of the combination of APCV with MCM and communicating hydrocephalus have been reported, but no mention was made in these cases of port-wine nevi. The port-wine nevus is generally considered to be a benign congenital skin disorder unassociated with any central nervous or any systemic anomalies. This port-wine nevus is not the same skin anomaly that is seen in the Sturge-Weber syndrome.

This is the first family reported in which there is an association between port-wine nevus and mega cisterna magna. The size of the cisterna magna is variable, but comparison of these cases and those of Adam and Greenberg would justify classifying these cases as MCM. According to Adam and Greenberg, MCM would be exhibited in four of every 1000 CT scans. No two or more members of the same family were presented in the above series. The association in five members of the same family would be more than chance alone could dictate.

A syndrome is presented in which five affected members of a family, representing three generations, have glabella port-wine nevi and mega cisterna magna. Two of the male siblings have identical communicating hydrocephalus pictures with agenesis of the posterior cerebellar vermis. The syndrome probably is transmitted as an autosomal dominant, with incomplete expressivity in the lesser affected members. Alternatively, but less likely, would be two separate genetic syndromes, an autosomal dominant, expressed as glabella port-wine nevus and mega cisterna magna, and an x-linked recessive, expressed as APCV and communicating hydrocephalus.

References

Familial hydrocephalus and mega cisterna magna


*Address reprint requests to: Harvey R. Nova, M.D., 200 Engle Street, Englewood, New Jersey 07631.*